

SUPPLEMENTARY INFORMATION

Programmable editing of a target base in genomic DNA without double-stranded DNA cleavage

Alexis C. Komor^{1,2}, Yongjoo B. Kim^{1,2}, Michael S. Packer^{1,2}, John A. Zuris^{1,2}, David R. Liu^{1,2*}

¹ Department of Chemistry and Chemical Biology, Harvard University, Cambridge, MA, 02138

² Howard Hughes Medical Institute, Harvard University, Cambridge, MA, 02138

* Correspondence should be addressed to David R. Liu: drliu@fas.harvard.edu

Supplementary Discussion

Supplementary Notes: Base-calling and indel-identification Matlab scripts.

Supplementary Sequences: Primer, gene, and protein sequences used in this work.

Supplementary Table 1: Activities of BE1, BE2, and BE3 at EMX1 off-targets.

Supplementary Table 2: Activities of BE1, BE2, and BE3 at FANCF off-targets.

Supplementary Table 3: Activities of BE1, BE2, and BE3 at HEK293 site 2 off-targets.

Supplementary Table 4: Activities of BE1, BE2, and BE3 at HEK293 site 3 off-targets.

Supplementary Table 5: Activities of BE1, BE2, and BE3 at HEK293 site 4 off-targets.

Supplementary Table 6: Indel formation following treatment of HEK293T cells with BE1, BE2, BE3, and wt Cas9 + a single-stranded DNA template for HDR.

Supplementary Table 7: Mutation rates of non-protospacer bases following BE3-mediated correction of the Alzheimer's disease-associated *APOE4* allele to *APOE3r* in mouse astrocytes.

Supplementary Table 8: Mutation rates of non-protospacer bases following BE3-mediated correction of the cancer-associated p53 Y163C mutation in HCC1954 human cells.

Supplementary Table 9: List of base-editable gene variants associated with human disease containing only a single C within the activity window and an NGG PAM positioned appropriately

Supplementary References

SUPPLEMENTARY DISCUSSION

Recent progress to increase HDR and suppress NHEJ. A small-molecule inhibitor of ligase IV, an essential enzyme in the NHEJ pathway, has been shown to increase HDR efficiency.^{35,36} However, this strategy is challenging in post-mitotic cells, which typically down-regulate HDR, and its therapeutic relevance is limited by the potential risks of inhibiting ligase IV in non-target cells. Enhanced HDR efficiency can also be achieved by the timed delivery of Cas9-guide RNA complexes into chemically synchronized cells, as HDR efficiency is highly cell-cycle dependent.³⁷ Such an approach, however, is limited to research applications in cell culture since synchronizing cells is highly disruptive. In some cases, it is possible to design HDR templates such that the product of successful HDR contains mutations in the PAM sequence and therefore is no longer a substrate for subsequent Cas9 modification, increasing the overall yield of HDR products,³⁸ although such an approach imposes constraints on the product sequences. Recently, this strategy has been coupled to the use of ssDNA donors that are complementary to the non-target strand and high-efficiency ribonucleoprotein (RNP) delivery to substantially increase the efficiency of HDR, but even in these cases the ratio of HDR to NHEJ outcomes is relatively low (< 2).³⁹

BE1 activity window. We observed some base editing outside of the typical window of positions 4 to 8 when the substrate C is preceded by a T, which we attribute to the unusually high activity of APOBEC1 for TC substrates.⁴⁰

Testing BE1 in human cells. To test base editing in human cells, we optimized BE1 codon usage for mammalian expression and appended a C-terminal nuclear localization signal (NLS).⁴¹

Indels in BE1- and BE2-treated cells. Indels will on rare occasion arise from the processing of U:G lesions by cellular repair processes, which involve single-strand break intermediates that are known to lead to indels.⁴² Given that several hundred endogenous U:G lesions are generated every day per human cell from spontaneous cytosine deamination,⁴³ we anticipate that the total indel frequency from U:G lesion repair is unlikely to increase from BE1 or BE2 activity at a single target locus.

Permanence of base editing in cells. We observed no substantial change in editing efficiency between non-passaged HEK293T cells (editing observed in 1.8% to 2.6% of sequenced strands

for the three target Cs with BE2, and 6.2% to 14.3% with BE3) and cells that had undergone approximately five cell divisions after base editing (editing observed in 1.9% to 2.3% of sequenced strands for the same target Cs with BE2, and 6.4% to 14.5% with BE3), confirming that base edits in these cells are durable (Extended Data Fig. 6).

Off-target activity. The off-target activities of Cas9, dCas9, and Cas9 nickase have been extensively studied.⁴⁴⁻⁴⁷ In general, off-target C to T conversions by BE1, BE2, and BE3 paralleled off-target Cas9 nuclease-mediated genome modification frequencies.

Determination of clinically relevant human genetic diseases treatable by BE2 and BE3. To illuminate the potential relevance of base editors to address human genetic diseases, we searched the NCBI ClinVar database⁴⁸ for known genetic diseases that could in principle be corrected by this approach. We filtered ClinVar by first examining only single nucleotide polymorphisms (SNPs), then removing any non-pathogenic variants. Out of the 24,670 pathogenic SNPs, 3,956 are caused by either a T to C, or an A to G, substitution. This list was further filtered to only include variants with a nearby NGG PAM that would position the SNP within the deamination activity window, resulting in 911 clinically relevant pathogenic gene variants that could in principle be corrected by the base editors described here. Of these, 284 contain only one C within the base editing activity window. A detailed list of these pathogenic mutations can be found in Supplementary Table 9.

SUPPLEMENTARY NOTES

Base Calling Matlab Script

```
WTnuc='GCGGACATGGAGGACGTGCGCGGCCGCCTGGTGCAGTACCGCGGCGAGGTGCAGGCCATGCTCGGCCAGA
GCACCGAGGAGCTGCGGGTGCCTCGCCTCCCACCTGCGCAAGCTGCGTAAGCGGCTCCTCCGGATGCCGATGAC
CTGCAGAACGCCCTGGCAGTGTACCAGGCCGGGCCGAGGGCGCCGAGCGCGGCCCTAGCGCCATCCCGAGCG
CCTGGGGCCCTGGTGGAACAG';
%cycle through fastq files for different samples
files=dir('*.fastq');
for d=1:20
    filename=files(d).name;
    %read fastq file
    [header,seqs,qscore] = fastqread(filename);
    seqsLength = length(seqs); % number of sequences
    seqsFile = strrep(filename,'.fastq','');
    % creates a directory with the same name as fastq file
    if exist(seqsFile,'dir');
        error('Directory already exists. Please rename or move it before
moving on.');
    end
    mkdir(seqsFile); % make directory
    wtLength = length(WTnuc); % length of wildtype sequence
    %% aligning back to the wildtype nucleotide sequence
    %
    % AlN is a matrix of the nucleotide alignment
    window=1:wtLength;
    sBLength = length(seqs); % number of sequences
    % counts number of skips
    nSkips = 0;
    ALN=repmat(' ',[sBLength wtLength]);
    % iterate through each sequencing read
    for i = 1:sBLength
        %If you only have forward read fastq files leave as is
        %If you have R1 foward and R2 is reverse fastq files uncomment the
        %next four lines of code and the subsequent end statement
        %
        if mod(d,2)==0;
        %
            reverse = seqrcomplement(seqs{i});
            %
            [score,alignment,start] =
            swalign(reverse,WTnuc,'Alphabet','NT');
        %
        else

            [score,alignment,start] = swalign(seqs{i},WTnuc,'Alphabet','NT');

        end

        % length of the sequencing read
        len = length(alignment(3,:));
        % if there is a gap in the alignment , skip = 1 and we will
        % throw away the entire read
        skip = 0;
        for j = 1:len
            if (alignment(3,j) == '-' || alignment(1,j) == '-')
                skip = 1;
                break;
            end
            %in addition if the qscore for any given base in the read is
            %below 31 the nucleotide is turned into an N (fastq qscores that
            are not letters)
        end
    end
end
```

```

        if isletter(qscore{i}(start(1)+j-1))
    else
        alignment(1,j) = 'N';
    end

    end
    if skip == 0 && len>10
        ALN(i, start(2):(start(2)+length(alignment)-1))=alignment(1,:);
    end
end
% with the alignment matrices we can simply tally up the occurrences of
% each nucleotide at each column in the alignment these
% tallies ignore bases annotated as N
% due to low qscores
TallyNTD=zeros(5,wtLength);
for i=1:wtLength

TallyNTD(:,i)=[sum(ALN(:,i)=='A'),sum(ALN(:,i)=='C'),sum(ALN(:,i)=='G'),sum(ALN(:,i)=='T'),sum(ALN(:,i)=='N')];
end
% we then save these tally matrices in the respective folder for
% further processing

save(strcat(seqsFile, '/TallyNTD'), 'TallyNTD');
dlmwrite(strcat(seqsFile, '/TallyNTD.txt'), TallyNTD, 'precision',
'%.3f', 'newline', 'pc');
end

```

INDEL Detection Matlab Script

```

WTnuc='GCGGACATGGAGGACGTGCGCGGCCGCCTGGTGCAGTACCGCGGCGAGGTGCAGGCCATGCTCGGCCAGA
GCACCGAGGAGCTGCGGGTGCCTCGCCTCCCACCTGCGCAAGCTGCGTAAGCGGCTCCTCCGCGATGCCGATGAC
CTGCAGAACGCCCTGGCAGTGTACCAGGCCGGGCCCGAGGGCGCCGAGCGCAGGCCATCCGCGAGCG
CCTGGGGCCCTGGTGGAACAG';
%cycle through fastq files for different samples
files=dir('*.fastq');
%specify start and width of indel window as well as length of each flank
indelstart=154;
width=30;
flank=10;

for d=1:3
    filename=files(d).name;
    %read fastq file
    [header,seqs,qscore] = fastqread(filename);
    seqsLength = length(seqs); % number of sequences
    seqsFile = strcat(strrep(filename,'.fastq',''), '_INDELS');
    %create a directory with the same name as fastq file+_INDELS
    if exist(seqsFile,'dir');
        error('Directory already exists. Please rename or move it before
moving on.');
    end
    mkdir(seqsFile); % make directory
    wtLength = length(WTnuc); % length of wildtype sequence
    sBLength = length(seqs); % number of sequences

    % initialize counters and cell arrays

```

```

nSkips = 0;
notINDEL=0;
ins={};
dels={};
NumIns=0;
NumDels=0;
% iterate through each sequencing read
for i = 1:sBLength
%search for 10BP sequences that should flank both sides of the "INDEL
WINDOW"
    windowstart=strfind(seqs{i},WTnuc(indelstart-flank:indelstart));

    windowend=strfind(seqs{i},WTnuc(indelstart+width:indelstart+width+flank
));
    %if the flanks are found proceed
    if length(windowstart)==1 && length(windowend)==1
        %if the sequence length matches the INDEL window length save as
        %not INDEL
        if windowend-windowstart==width+flank
            notINDEL=notINDEL+1;
        %if the sequence is two or more bases longer than the INDEL
        %window length save as an Insertion
        elseif windowend-windowstart>=width+flank+2
            NumIns=NumIns+1;
            ins{NumIns}=seqs{i};
        %if the sequence is two or more bases shorter than the INDEL
        %window length save as a Deletion
        elseif windowend-windowstart<=width+flank-2
            NumDels=NumDels+1;
            dels{NumDels}=seqs{i};
        %keep track of skipped sequences that are either one base
        %shorter or longer than the INDEL window width
        else
            nSkips=nSkips+1;
        end
    %keep track of skipped sequences that do not possess matching flank
    %sequences
    else
        nSkips=nSkips+1;
    end
end

fid=fopen(strcat(seqsFile, '/summary.txt'), 'wt');
fprintf(fid, 'Skipped reads %i\n not INDEL %i\n Insertions %i\n Deletions
%i\n', [nSkips, notINDEL, NumIns, NumDels]);
fclose(fid);
save(strcat(seqsFile, '/nSkips'), 'nSkips');
save(strcat(seqsFile, '/notINDEL'), 'notINDEL');
save(strcat(seqsFile, '/NumIns'), 'NumIns');
save(strcat(seqsFile, '/NumDels'), 'NumDels');
save(strcat(seqsFile, '/dels'), 'dels');
C = dels;
fid = fopen(strcat(seqsFile, '/dels.txt'), 'wt');
fprintf(fid, '%s\n', C{:});
fclose(fid);
save(strcat(seqsFile, '/ins'), 'ins');
C = ins;

```

```
fid = fopen(strcat(seqsFile, '/ins.txt'), 'wt');
fprintf(fid, '"%s"\n', C{::});
fclose(fid);

end
```

SUPPLEMENTARY SEQUENCES

All oligonucleotides were purchased from Integrated DNA Technologies (IDT).

Primers used for generating sgRNA transfection plasmids. rev_sgRNA_plasmid was used in all cases. The pFYF1320 plasmid was used as template as noted in Methods section.

rev_sgRNA_plasmid	GGTGTTCGTCCTTCCACAAG
fwd_p53_Y163C	GCTTGCAGATGCCATGGCGGTTTAGAGCTAGAAATAGCAAGTAAAATAAGGC
fwd_APOE4_C158R	GAAGCGCCTGGCAGTGTACCGTTAGAGCTAGAAATAGCAAGTAAAATAAGGC
fwd_EMX1	GAGTCCGAGCAGAAGAAGAAGTTAGAGCTAGAAATAGCAAGTAAAATAAGGC
fwd_FANCF	GGAATCCCTCTGCAGCACCGTTAGAGCTAGAAATAGCAAGTAAAATAAGGC
fwd_HEK293_2	GAACACAAAGCATAGACTCGTTAGAGCTAGAAATAGCAAGTAAAATAAGGC
fwd_HEK293_3	GGCCCAGACTGAGCACGTGAGTTAGAGCTAGAAATAGCAAGTAAAATAAGGC
fwd_HEK293_4	GGCACTGCGGCTGGAGGTGGTTAGAGCTAGAAATAGCAAGTAAAATAAGGC
fwd_RNF2	GTCATCTTAGTCATTACCTGGTTAGAGCTAGAAATAGCAAGTAAAATAAGGC

Sequences of all ssDNA substrates used in *in vitro* deaminase assays.

rAPOBEC1 substrate	Cy3-ATTATTATTATTCCGGGATTATTATTATTATTATTATT
hAID/pmCDA1 substrate	Cy3-ATTATTATTATTAGCTATTATTATTATTATTATT
hAPOBEC3G substrate	Cy3-ATTATTATTATCCGGATTATTATTATTATTATT

Primers used for generating PCR products to serve as substrates for T7 transcription of sgRNAs for gel-based deaminase assay. rev_gRNA_T7 was used in all cases. The pFYF1320 plasmid was used as template as noted in Methods section.

rev_sgRNA_T7	AAAAAAAGCACCGACTCGGTG
fwd_sgRNA_T7_dsDNA_2	TAATACGACTCACTATAGGCCGGATTATTATTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_3	TAATACGACTCACTATAGGTCCGGGATTATTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_4	TAATACGACTCACTATAGGTCCGGGATTATTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_5	TAATACGACTCACTATAGGATTCCGGGATTATTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_6	TAATACGACTCACTATAGGTATTCCGGGATTATTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_7	TAATACGACTCACTATAGGTATTCCGGGATTATTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_8	TAATACGACTCACTATAGGATTATTCCGGGATTATTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_9	TAATACGACTCACTATAGGTATTCCGGGATTATTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_10	TAATACGACTCACTATAGGATTATTCCGGGATTATTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_11	TAATACGACTCACTATAGGTATTATTCCGGGATTATTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_12	TAATACGACTCACTATAGGTATTATTCCGGGATTATTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_13	TAATACGACTCACTATAGGATTATTCCGGGATTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_14	TAATACGACTCACTATAGGTATTATTCCGGGATTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_15	TAATACGACTCACTATAGGATTATTACCGGGAGTTAGAGCTAGAAATAGCA

fwd_sgRNA_T7_dsDNA_18	TAATACGACTCACTATAGGATTATTATTACCGCGTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_noC	TAATACGACTCACTATAGGATATTAATTATTAAAGTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_APOE4_C112R	TAATACGACTCACTATAGGGAGGACGTGCGCGCCGCCGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_APOE4_C158R	TAATACGACTCACTATAGGAAGCGCCTGGCAGTGTACCGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_CTNNB1_T41A	TAATACGACTCACTATAGGCTGTGGCAGTGGACCAGAAGTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_HRAS_Q61R	TAATACGACTCACTATAGGCCTCCGGCCGGTATCCGTTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_53_Y163C	TAATACGACTCACTATAGGGCTTGAGATGCCATGGCGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_53_Y236C	TAATACGACTCACTATAGGACACATGCAGTTGAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_dsDNA_53_N239D	TAATACGACTCACTATAGGTGTCACACATGTAGTTAGGTTAGAGCTAGAAATAGCA

Sequences of 80-nucleotide unlabeled strands and Cy3-labeled universal primer used in gel-based dsDNA deaminase assays.

Cy3-primer	Cy3-GTAGGTAGTTAGGATGAATGGAAGGGTGGTA
dsDNA_2	GTCATGGATCCAGAGGTATCCATTAAATAAAATCCGGGGTATACCAACCTCCATTATCCTAACTACCTAC
dsDNA_3	GTCATGGATCCAGAGGTATCCATAAAATAAAATCCGCGGAAGCTATACCAACCTCCATTATCCTAACTACCTAC
dsDNA_4	GTCATGGATCCAGAGGTATCCATAAAATAAAATCCGCGGAAGGTATACCAACCTCCATTATCCTAACTACCTAC
dsDNA_5	GTCATGGATCCAGAGGTATCCAATAAAATAATCCGCGGAATGGCTATACCAACCTCCATTATCCTAACTACCTAC
dsDNA_6	GTCATGGATCCAGAGGTATCCAATAAAATAATCCGCGGAATAGGCTATACCAACCTCCATTATCCTAACTACCTAC
dsDNA_7	GTCATGGATCCAGAGGTATCCATAAAATAATCCGCGGAATAAGGCTATACCAACCTCCATTATCCTAACTACCTAC
dsDNA_8	GTCATGGATCCAGAGGTATCCAATAAAATAATCCGCGGAATAATGGCTATACCAACCTCCATTATCCTAACTACCTAC
dsDNA_9	GTCATGGATCCAGAGGTATCCAATAAAATCCGCGGAATAATAGGCTATACCAACCTCCATTATCCTAACTACCTAC
dsDNA_10	GTCATGGATCCAGAGGTATCCAATAAAATCCGCGGATAATAATGGCTATACCAACCTCCATTATCCTAACTACCTAC
dsDNA_11	GTCATGGATCCAGAGGTATCCATAAAATCCGCGGAATATAATAGGCTATACCAACCTCCATTATCCTAACTACCTAC
dsDNA_12	GTCATGGATCCAGAGGTATCCAATAAAATCCGCGGAATATAAGGCTATACCAACCTCCATTATCCTAACTACCTAC
dsDNA_13	GTCATGGATCCAGAGGTATCCAATAAAATCCGCGGAATATAATAGGCTATACCAACCTCCATTATCCTAACTACCTAC
dsDNA_14	GTCATGGATCCAGAGGTATCCAATCCGCGGAATATAATAATAGGCTATACCAACCTCCATTATCCTAACTACCTAC
dsDNA_15	GTCATGGATCCAGAGGTATCCATCCGCGGATAATAATAATGGCTATACCAACCTCCATTATCCTAACTACCTAC
dsDNA_18	GTCATGGATCCAGAGGTATCCAGCGGTATAATAATAATGGCTATACCAACCTCCATTATCCTAACTACCTAC
dsDNA_noC	GTCATGGATCCAGAGGTATCCATTAAATAAAATAATTAAATTACTATACCAACCTCCATTATCCTAACTACCTAC
dsDNA_8U	5Cy3-GTAGGTAGTTAGGATGAATGGAAGGGTGGTAGATTATTACUGCGGATTATTGGATGACCTCTGGATCCATGGACAT
dsDNA_APOE_C112R	GCACCTCGCCCGGGTACTGCACCAGGCGGCCGCGCACGTCTCCATGTCTACCAACCTCCATTATCCTAACTACCTAC
dsDNA_APOE_C158R	CGGCGCCCTCGCGGGCCCCGGCTGGTACACTGCCAGCGCTCTGCAGTACCAACCTCCATTATCCTAACTACCTAC
dsDNA_CTNNB1_T41A	GTCTTACCTGGACTCTGAATCCATTCTGGTCCACTGCCACAGCTCTTACCAACCTCCATTATCCTAACTACCTAC
dsDNA_HRAS_Q61R	GGAGACGTGCCTGTTGGACATCCTGGATACCGCCGGGGAGGAGTACTACCAACCTCCATTATCCTAACTACCTAC
dsDNA_p53_Y163C	ACCCCCGCCCCGCACCCGCGTCCCGCCCATGGCCTGCAAGCAGTCATACCAACCTCCATTATCCTAACTACCTAC
dsDNA_p53_Y236C	AGGTTGGCTCTGACTGTACCAACCACCAACTACAATGCTGTAACAGTACCAACCTCCATTATCCTAACTACCTAC
dsDNA_p53_N239D	TGGCTCTGACTGTACCAACCACCAACTACAATGCTGTAACAGTACCAACCTCCATTATCCTAACTACCTAC

Primers used for generating PCR products to serve as substrates for T7 transcription of sgRNAs for high-throughput sequencing. rev_gRNA_T7 (above) was used in all cases. The pFYF1320 plasmid was used as template as noted in Methods section.

fwd_sgRNA_T7_HTS_base	TAATACGACTCACTATAGGTTATTCGTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_1A	TAATACGACTCACTATAGGATATTCGTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_1C	TAATACGACTCACTATAGGCATATTCGTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_1G	TAATACGACTCACTATAGGTATTCGTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_2A	TAATACGACTCACTATAGGAATATTCGTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_2C	TAATACGACTCACTATAGGCATTTCGTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_2G	TAATACGACTCACTATAGGTATTCGTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_3T	TAATACGACTCACTATAGGTTTTCGTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_3C	TAATACGACTCACTATAGGTTCTTCGTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_3G	TAATACGACTCACTATAGGTTCTCGTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_4A	TAATACGACTCACTATAGGTAAATCGTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_4C	TAATACGACTCACTATAGGTTACTCGTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_4G	TAATACGACTCACTATAGGTAGTCGTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_5A	TAATACGACTCACTATAGGTATATCGTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_5C	TAATACGACTCACTATAGGTATCTCGTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_5G	TAATACGACTCACTATAGGTATGTCGTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_6A	TAATACGACTCACTATAGGTATTACGTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_6C	TAATACGACTCACTATAGGTATTCCGTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_6G	TAATACGACTCACTATAGGTATTGCGTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_8A	TAATACGACTCACTATAGGTATTCATGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_8T	TAATACGACTCACTATAGGTATTCCTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_8C	TAATACGACTCACTATAGGTATTCCGTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_9A	TAATACGACTCACTATAGGTATTCGAGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_9C	TAATACGACTCACTATAGGTATTCGCGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_9G	TAATACGACTCACTATAGGTATTCGGGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_10A	TAATACGACTCACTATAGGTATTCGTAGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_10T	TAATACGACTCACTATAGGTATTCGTGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_10C	TAATACGACTCACTATAGGTATTCGTGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_11A	TAATACGACTCACTATAGGTATTCGTGAATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_11T	TAATACGACTCACTATAGGTATTCGTGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_11C	TAATACGACTCACTATAGGTATTCGTGCATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_12T	TAATACGACTCACTATAGGTATTCGTGGTTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_12C	TAATACGACTCACTATAGGTATTCGTGGCTTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_12G	TAATACGACTCACTATAGGTATTCGTGGGTTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_13A	TAATACGACTCACTATAGGTATTCGTGGAATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_13C	TAATACGACTCACTATAGGTATTCGTGGACTTATTTAGTTAGAGCTAGAAATAGCA

fwd_sgRNA_T7_HTS_13G	TAATACGACTCACTATAGGTATTCTGGAGTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_multiC	TAATACGACTCACTATAGGTCCCCCCCCGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_TCGCACCC_odd	TAATACGACTCACTATAGGCACCCGTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_CCTCGCAC_odd	TAATACGACTCACTATAGGCTCGCACGTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ACCCTCGC_odd	TAATACGACTCACTATAGGCCCTCGCGTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_GCACCCTC_odd	TAATACGACTCACTATAGGCACCCCTCGCGTGGATTATTTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_TCGCACCC_even	TAATACGACTCACTATAGGTCGACCCGTGGATTATTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_CCTCGCAC_even	TAATACGACTCACTATAGGCCCTCGCACGTGGATTATTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_ACCCTCGC_even	TAATACGACTCACTATAGGACCCCTCGCGTGGATTATTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_GCACCCTC_even	TAATACGACTCACTATAGGGCACCCCTCGTGATTATTAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_EMX1	TAATACGACTCACTATAGGAGTCGAGCAGAAGAAGAAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_FANCF	TAATACGACTCACTATAGGGAATCCCTCTGCAGCACCGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_HEK293_site2	TAATACGACTCACTATAGGAACACAAGCATAGACTGCAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_HEK293_site3	TAATACGACTCACTATAGGGCCCAGACTGAGCACGTGAGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_HEK293_site4	TAATACGACTCACTATAGGGCACTGCGCTGGAGGTGGTTAGAGCTAGAAATAGCA
fwd_sgRNA_T7_HTS_RNF2	TAATACGACTCACTATAGGTCATCTAGCATTACCTGGTTAGAGCTAGAAATAGCA

Sequences of *in vitro*-edited dsDNA for high-throughput sequencing (HTS). Shown are the sequences of edited strands. Reverse complements of all sequences shown were also obtained. dsDNA substrates were obtained by annealing complementary strands as described in Methods. Oligonucleotides representing the EMX1, FANCF, HEK293 site 2, HEK293 site 3, HEK293 site 4, and RNF2 loci were originally designed for use in the gel-based deaminase assay and therefore have the same 25-nt sequence on their 5'- ends (matching that of the Cy3-primer).

Base sequence	ACGTAAACGGCCACAAGTTCTTATTCGTGGATTATTATGGCATCTTCAAGGACG
1A	ACGTAAACGGCCACAAGTTCATATTCGTGGATTATTATGGCATCTTCAAGGACG
1C	ACGTAAACGGCCACAAGTTCTATTCGTGGATTATTATGGCATCTTCAAGGACG
1G	ACGTAAACGGCCACAAGTTGTATTCGTGGATTATTATGGCATCTTCAAGGACG
2A	ACGTAAACGGCCACAAGTTCTAATTCGTGGATTATTATGGCATCTTCAAGGACG
2C	ACGTAAACGGCCACAAGTTCTATTCGTGGATTATTATGGCATCTTCAAGGACG
2G	ACGTAAACGGCCACAAGTTCTGATTCGTGGATTATTATGGCATCTTCAAGGACG
3T	ACGTAAACGGCCACAAGTTCTTTCTGTGGATTATTATGGCATCTTCAAGGACG

3C ACGTAAACGGCCACAAGTTCTTCGTGGATTATTTATGGCATCTTCAAGGACG
3G ACGTAAACGGCCACAAGTTCTTCGTGGATTATTTATGGCATCTTCAAGGACG
4A ACGTAAACGGCCACAAGTTCTTAATCGTGGATTATTTATGGCATCTTCAAGGACG
4C ACGTAAACGGCCACAAGTTCTACTCGTGGATTATTTATGGCATCTTCAAGGACG
4G ACGTAAACGGCCACAAGTTCTAGTCGTGGATTATTTATGGCATCTTCAAGGACG
5A ACGTAAACGGCCACAAGTTCTATATCGTGGATTATTTATGGCATCTTCAAGGACG
5C ACGTAAACGGCCACAAGTTCTATCTCGTGGATTATTTATGGCATCTTCAAGGACG
5G ACGTAAACGGCCACAAGTTCTATGCGTGGATTATTTATGGCATCTTCAAGGACG
6A ACGTAAACGGCCACAAGTTCTTACGTGGATTATTTATGGCATCTTCAAGGACG
6C ACGTAAACGGCCACAAGTTCTTATTCGTGGATTATTTATGGCATCTTCAAGGACG
6G ACGTAAACGGCCACAAGTTCTTATTGCGTGGATTATTTATGGCATCTTCAAGGACG
8A ACGTAAACGGCCACAAGTTCTTATTCATGGATTATTTATGGCATCTTCAAGGACG
8T ACGTAAACGGCCACAAGTTCTTATTCGTGGATTATTTATGGCATCTTCAAGGACG
8C ACGTAAACGGCCACAAGTTCTTATTCGTGGATTATTTATGGCATCTTCAAGGACG
9A ACGTAAACGGCCACAAGTTCTTATTCGAGGATTATTTATGGCATCTTCAAGGACG
9C ACGTAAACGGCCACAAGTTCTTATTCGCGGATTATTTATGGCATCTTCAAGGACG
9G ACGTAAACGGCCACAAGTTCTTATTCGGGGATTATTTATGGCATCTTCAAGGACG
10A ACGTAAACGGCCACAAGTTCTTATTCGTAGATTATTTATGGCATCTTCAAGGACG
10T ACGTAAACGGCCACAAGTTCTTATTCGTGATTATTTATGGCATCTTCAAGGACG
10C ACGTAAACGGCCACAAGTTCTTATTCGTGATTATTTATGGCATCTTCAAGGACG
11A ACGTAAACGGCCACAAGTTCTTATTCGTGAATTATTTATGGCATCTTCAAGGACG
11T ACGTAAACGGCCACAAGTTCTTATTCGTGTATTATTTATGGCATCTTCAAGGACG
11C ACGTAAACGGCCACAAGTTCTTATTCGTGCATTATTTATGGCATCTTCAAGGACG
12T ACGTAAACGGCCACAAGTTCTTATTCGTGGTTATTATGGCATCTTCAAGGACG
12C ACGTAAACGGCCACAAGTTCTTATTCGTGGCTTATTATGGCATCTTCAAGGACG
12G ACGTAAACGGCCACAAGTTCTTATTCGTGGTTATTATGGCATCTTCAAGGACG
13A ACGTAAACGGCCACAAGTTCTTATTCGTGGAATTATTTATGGCATCTTCAAGGACG
13C ACGTAAACGGCCACAAGTTCTTATTCGTGGACTTATTATGGCATCTTCAAGGACG
13G ACGTAAACGGCCACAAGTTCTTATTCGTGGAGTTATTATGGCATCTTCAAGGACG
multiC ACGTAAACGGCCACAAGTTCTTCCCCCCCCGATTATTTATGGCATCTTCAAGGACG
TCGCACCC_odd ACGTAAACGGCCACAAGTTCGCACCCGTGGATTATTTATGGCATCTTCAAGGACG
CCTCGCAC_odd ACGTAAACGGCCACAAGTTCTCGCACGTGGATTATTTATGGCATCTTCAAGGACG
ACCCTCGC_odd ACGTAAACGGCCACAAGTTACCCCTCGCGTGGATTATTTATGGCATCTTCAAGGACG
GCACCCCTC_odd ACGTAAACGGCCACAAGTTGCACCCCTCGTGGATTATTTATGGCATCTTCAAGGACG
TCGCACCC_even ACGTAAACGGCCACAAGTATCGCACCCGTGGATTATTTATGGCATCTTCAAGGACG
CCTCGCAC_even ACGTAAACGGCCACAAGTATCCTCGCACGTGGATTATTTATGGCATCTTCAAGGACG
ACCCTCGC_even ACGTAAACGGCCACAAGTACCCCTCGCGTGGATTATTTATGGCATCTTCAAGGACG
GCACCCCTC_even ACGTAAACGGCCACAAGTATGCACCCCTCGTGGATTATTTATGGCATCTTCAAGGACG

EMX1_invitro	GTAGGTAGTTAGGATGAATGGAAGGTTGGTAGGCCTGAGTCGAGCAGAAGAAGAAGGGCTCCCATCACATCAACCGGTG
FANCF_invitro	GTAGGTAGTTAGGATGAATGGAAGGTTGGTACTCATGGAATCCCTCTGCAGCACCTGGATCGCTTCCGAGCTCTGG
HEK293_site2_invitro	GTAGGTAGTTAGGATGAATGGAAGGTTGGAAACTGGAACACAAAGCATAGACTGCAGGGCAGCCTGAATAGCTG
HEK293_site3_invitro	GTAGGTAGTTAGGATGAATGGAAGGTTGGTACTGGGGCCCAGACTGAGCACGTGATGGCAGAGGAAGGAAGCCCTGCT
HEK293_site4_invitro	GTAGGTAGTTAGGATGAATGGAAGGTTGGTACCGTGGCACTGCAGGGCTGGAGGTGGGGTAAAGCGGAGACTCTGGTGC
RNF2_invitro	GTAGGTAGTTAGGATGAATGGAAGGTTGGTATGGCAGTCATCTTAGTCATTACCTGAGGTGGTGTAACTCATATAA

Primers for HTS of *in vitro* edited dsDNA.

fwd_invitroHTS	ACACTCTTCCCTACACGACGCTTCCGATCTNNNNACGTAAACGGCCACAA
rev_invitroHTS	TGGAGTTCAGACGTGTGCTCTCCGATCTCGTCTTGAGAAGAAGATGC
fwd_invitro_HEK_targets	ACACTCTTCCCTACACGACGCTTCCGATCTNNNNTAGGTAGTTAGGATGAATGGAA
rev_EMX1_invitro	TGGAGTTCAGACGTGTGCTCTCCGATCTACCGGTTGATGTGATGG
rev_FANCF_invitro	TGGAGTTCAGACGTGTGCTCTCCGATCTCAGAAAGCTCGGAAAAGC
rev_HEK293_site2_invitro	TGGAGTTCAGACGTGTGCTCTCCGATCTCAGCTATTCAAGGCTGGC
rev_HEK293_site3_invitro	TGGAGTTCAGACGTGTGCTCTCCGATCTAGCAGGGCTTCCTTC
rev_HEK293_site4_invitro	TGGAGTTCAGACGTGTGCTCTCCGATCTGCACCAGAGTCTCCG
rev_RNF2_invitro	TGGAGTTCAGACGTGTGCTCTCCGATCTTATGAGTTACAACGAACACC

Primers for HTS of on-target and off-target sites from all mammalian cell culture experiments.

fwd_EMX1HTS	ACACTCTTCCCTACACGACGCTTCCGATCTNNNNACGCTAGCCTGAGTGTGA
rev_EMX1HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTCTCGTGGTTGTGGTTGC
fwd_FANCFHTS	ACACTCTTCCCTACACGACGCTTCCGATCTNNNNCATTGAGAGAGGCATATCA
rev_FANCFHTS	TGGAGTTCAGACGTGTGCTCTCCGATCTGGGTCCCAGGTGCTGAC
fwd_HEK293_site2HTS	ACACTCTTCCCTACACGACGCTTCCGATCTNNNNCAGCCCCATCTGTCAAAC
rev_HEK293_site2HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTTGAATGGATTCTTGAAACAATGA
fwd_HEK293_site3HTS	ACACTCTTCCCTACACGACGCTTCCGATCTNNNNATGTGGGCTGCCTAGAAAGG
rev_HEK293_site3HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTCCAGCCAACCTGTCAACC
fwd_HEK293_site4HTS	ACACTCTTCCCTACACGACGCTTCCGATCTNNNNGAACCCAGGTAGCCAGAGAC
rev_HEK293_site4HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTTCAACCCGAACGGAG
fwd_RNF2HTS	ACACTCTTCCCTACACGACGCTTCCGATCTNNNNACGCTCATATGCCCTTGG
rev_RNF2HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTACGTAGGAATTGGTGGGACA
fwd_p53_Y163CHTS	ACACTCTTCCCTACACGACGCTTCCGATCTNNNTGCCCTGACTTTCAACTCTGT
rev_p53_Y163CHTS	TGGAGTTCAGACGTGTGCTCTCCGATCTCAACCAGCCCTGCGTCTC
fwd_APOE4_C158RHTS	ACACTCTTCCCTACACGACGCTTCCGATCTNNNNGGACATGGAGGACGTGCG
rev_APOE4_C158RHTS	TGGAGTTCAGACGTGTGCTCTCCGATCTCTCCGTAGCGGCTGGCC
fwd_EMX1_off1HTS	ACACTCTTCCCTACACGACGCTTCCGATCTNNNNNTGCCCAATCATTGATGCTTT
rev_EMX1_off1HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTAGAAACATTACCATAGACTATCACCT

fwd_EMX1_off2HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNNAGTAGCCTCTTCATGTGC
rev_EMX1_off2HTS	TGGAGTTAGACGTGTGCTTCCGATCTGCTTCACAAGGATGCAGTCT
fwd_EMX1_off3HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNNAGACTAGACTCCGAGGGGA
rev_EMX1_off3HTS	TGGAGTTAGACGTGTGCTTCCGATCTTCCCGTCTGCTCACTT
fwd_EMX1_off4HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNNAGAGGCTGAAGAGGAAGACCA
rev_EMX1_off4HTS	TGGAGTTAGACGTGTGCTTCCGATCTGGCCCAGCTGTGCATTCTAT
fwd_EMX1_off6HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNNCCAAGAGGGCCAAGTCCTG
rev_EMX1_off6HTS	TGGAGTTAGACGTGTGCTTCCGATCTCAGCGAGGAGTGACAGCC
fwd_EMX1_off7HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNNCACTCCACCTGATCTCGGGG
rev_EMX1_off7HTS	TGGAGTTAGACGTGTGCTTCCGATCTCGAGGAGGGAGGGAGCAG
fwd_EMX1_off8HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNNACCACAAATGCCAAGAGAC
rev_EMX1_off8HTS	TGGAGTTAGACGTGTGCTTCCGATCTGACACAGTCAAGGGCCGG
fwd_EMX1_off9HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNNCCACCTTGAGGAGGCAA
rev_EMX1_off9HTS	TGGAGTTAGACGTGTGCTTCCGATCTTCCATCTGAGAAGAGAGTGGT
fwd_EMX1_off10HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNNNGTACACCTGGCCCTTCCT
rev_EMX1_off10HTS	TGGAGTTAGACGTGTGCTTCCGATCTTCCCTAGGCCACACCAG
fwd_FANCF_off1HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNNAAACCACTGAAGAACGCAGGG
rev_FANCF_off1HTS	TGGAGTTAGACGTGTGCTTCCGATCTGGTGCTTAATCCGGCTCCAT
fwd_FANCF_off2HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNNNTCCAGTGTTCATCCGAA
rev_FANCF_off2HTS	TGGAGTTAGACGTGTGCTTCCGATCTCCTCTGACCTCCACAACACTCT
fwd_FANCF_off3HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNNCTGGGTACAGTTGCGTGT
rev_FANCF_off3HTS	TGGAGTTAGACGTGTGCTTCCGATCTTCACTCTGAGCATGCCAAG
fwd_FANCF_off4HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNNNGTTAGAGCCAGTGAACTAGAG
rev_FANCF_off4HTS	TGGAGTTAGACGTGTGCTTCCGATCTGCAAGACAAATCCTTTACTTTG
fwd_FANCF_off5HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNNGGAGGGACGGCCTTAC
rev_FANCF_off5HTS	TGGAGTTAGACGTGTGCTTCCGATCTGCCCTGGCGAACATGGC
fwd_FANCF_off6HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNNCTGGTTAAGAGCATGGC
rev_FANCF_off6HTS	TGGAGTTAGACGTGTGCTTCCGATCTGATTGAGTCCCCACAGCACA
fwd_FANCF_off7HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNNCCAGTGTTCATCCCCAA
rev_FANCF_off7HTS	TGGAGTTAGACGTGTGCTTCCGATCTTGACCTCCACAACCTGGAAAAT
fwd_FANCF_off8HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNNNGCTCCAGACCCACCTGAAG
rev_FANCF_off8HTS	TGGAGTTAGACGTGTGCTTCCGATCTACCGAGGAAAATTGCTTGTG
fwd_HEK293_site2_off1HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNNNGTGGAGAGTGAGTAAGCCA
rev_HEK293_site2_off1HTS	TGGAGTTAGACGTGTGCTTCCGATCTACGGTAGGATGATTTCAGGCA
fwd_HEK293_site2_off2HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNNCACAAAGCAGTGTAGCTCAGG
rev_HEK293_site2_off2HTS	TGGAGTTAGACGTGTGCTTCCGATCTTTTGGTACTCGAGTGTATTCA
fwd_HEK2_ChIP_off1HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNNAGACAGGCTCAGGAAAGCTGT
rev_HEK2_ChIP_off1HTS	TGGAGTTAGACGTGTGCTTCCGATCTACACAAGCCTTCTCCAGGG
fwd_HEK2_ChIP_off2HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNAATAGGGGTGAGACTGGGG

rev_HEK2_ChIP_off2HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTGCCTCAGACGAGACTTGAGG
fwd_HEK2_ChIP_off3HTS	ACACTCTTCCCTACACGACGCTCTCCGATCTNNNNGGCCAGCAGGAAAGGAATCT
rev_HEK2_ChIP_off3HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTTGACTGCACCTGTAGCCATG
fwd_HEK2_ChIP_off4HTS	ACACTCTTCCCTACACGACGCTCTCCGATCTNNNTCAAGGAAATCACCCCTGCC
rev_HEK2_ChIP_off4HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTAACCTGGTGTGCAGCT
fwd_HEK2_ChIP_off5HTS	ACACTCTTCCCTACACGACGCTCTCCGATCTNNNATGGGCTCAGCTACGTCATG
rev_HEK2_ChIP_off5HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTAACAGTGTGGTGGCAA
fwd_HEK293_site3_off1HTS	ACACTCTTCCCTACACGACGCTCTCCGATCTNNNTCCCTGTTGACCTGGAGAA
rev_HEK293_site3_off1HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTCACTGACTTGCCTGACCA
fwd_HEK293_site3_off2HTS	ACACTCTTCCCTACACGACGCTCTCCGATCTNNNTGGTGTGACAGGGAGCAA
rev_HEK293_site3_off2HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTGAGATGTGGCAGAAGGG
fwd_HEK293_site3_off3HTS	ACACTCTTCCCTACACGACGCTCTCCGATCTNNNTGAGAGGAAACAGAAGGGCT
rev_HEK293_site3_off3HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTGTCAAAGGCCAAGAACCT
fwd_HEK293_site3_off4HTS	ACACTCTTCCCTACACGACGCTCTCCGATCTNNNTCTAGCACTTGGAAAGGTCG
rev_HEK293_site3_off4HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTGCTCATCTTAATCTGCTCAGCC
fwd_HEK293_site3_off5HTS	ACACTCTTCCCTACACGACGCTCTCCGATCTNNNAAGGAGCAGCTCTCCTGG
rev_HEK293_site3_off5HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTGTCTGACCACATCCCACAA
fwd_HEK3_ChIP_off1HTS	ACACTCTTCCCTACACGACGCTCTCCGATCTNNNCGCACATCCTTGTCTCT
rev_HEK3_ChIP_off1HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTACTGGAGCACCCCCAAG
fwd_HEK3_ChIP_off2HTS	ACACTCTTCCCTACACGACGCTCTCCGATCTNNNTGGTCACGTAGCTTGGC
rev_HEK3_ChIP_off2HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTGGTGGCCATGTGCAACTAA
fwd_HEK3_ChIP_off3HTS	ACACTCTTCCCTACACGACGCTCTCCGATCTNNNCTACTACGTGCCAGCTCAGG
rev_HEK3_ChIP_off3HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTACCTCCCCTCCACTAACCC
fwd_HEK3_ChIP_off4HTS	ACACTCTTCCCTACACGACGCTCTCCGATCTNNNGCCTCAGCTCCATTCTGT
rev_HEK3_ChIP_off4HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTAACCTTATGGCACCAGGGG
fwd_HEK3_ChIP_off5HTS	ACACTCTTCCCTACACGACGCTCTCCGATCTNNNGAGCTCAGCATTAGCAGGCT
rev_HEK3_ChIP_off5HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTTCCTGGTTCCGATTCCC
fwd_HEK293_site4_off1HTS	ACACTCTTCCCTACACGACGCTCTCCGATCTNNNGGATGGCTCTGAGACTCA
rev_HEK293_site4_off1HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTGTCTCCCTGCACCTCCGTCTTT
fwd_HEK293_site4_off2HTS	ACACTCTTCCCTACACGACGCTCTCCGATCTNNNTGGCAATGGAGGCATTGG
rev_HEK293_site4_off2HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTGAAGAGGCTGCCATGAGAG
fwd_HEK293_site4_off3HTS	ACACTCTTCCCTACACGACGCTCTCCGATCTNNNGGCTGAGGCTCGAACCTG
rev_HEK293_site4_off3HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTGTGGCCTCCATATCCCTG
fwd_HEK293_site4_off4HTS	ACACTCTTCCCTACACGACGCTCTCCGATCTNNNTTCCACCAGAACTCAGCCC
rev_HEK293_site4_off4HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTCCTCGGTTCCACAACAC
fwd_HEK293_site4_off5HTS	ACACTCTTCCCTACACGACGCTCTCCGATCTNNNCACGGGAAGGACAGGAGAAG
rev_HEK293_site4_off5HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTGAGGGAGGGATAAAGCAG
fwd_HEK293_site4_off6HTS	ACACTCTTCCCTACACGACGCTCTCCGATCTNNNCACGGGAGATGGCTTATGT
rev_HEK293_site4_off6HTS	TGGAGTTCAGACGTGTGCTCTCCGATCTCACATCCTCACTGTGCCACT

fwd_HEK293_site4_off7HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNNGTCAGTCTCGGGCCCTCA
rev_HEK293_site4_off7HTS	TGGAGTTAGACGTGTGCTCTCCGATCTGCCACTGTAAAGCTCTGGG
fwd_HEK293_site4_off8HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNAGGGTAGAGGGACAGAGCTG
rev_HEK293_site4_off8HTS	TGGAGTTAGACGTGTGCTCTCCGATCTGGACCCCACATAGTCAGTGC
fwd_HEK293_site4_off9HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNGCTGTCAGCCCATCTCCATC
rev_HEK293_site4_off9HTS	TGGAGTTAGACGTGTGCTCTCCGATCTGGGCAATTAGGACAGGGAC
fwd_HEK293_site4_off10HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNGCAGCGGAGGAGGTAGATTG
rev_HEK293_site4_off10HTS	TGGAGTTAGACGTGTGCTCTCCGATCTCTCAGTACCTGGAGTCCCCA
fwd_HEK4_ChIP_off1HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNGTCAATTGGAGGAGGAGCT
rev_HEK4_ChIP_off1HTS	TGGAGTTAGACGTGTGCTCTCCGATCTCACAGCAGAACACA
fwd_HEK4_ChIP_off3HTS	ACACTCTTCCCTACAGACGCTTCCGATCTNNNCCTACCCCAACACAGATGG
rev_HEK4_ChIP_off3HTS	TGGAGTTAGACGTGTGCTCTCCGATCTCACACA CAGGT CCTCC

Sequences of single-stranded oligonucleotide donor templates (ssODNs) used in HDR studies.

EMX1 sense

TCATCTGTGCCCTCCCTGGCCAGGTGAAGGTGTGGTCCAGAACCGGAGGACAAAGTACA
 AACGGCAGAAGCTGGAGGAGGAAGGGCCTGAGTTGAGCAGAACAGAAGGGCTCCATCACATC
 AACCGGTGGCGCATTGCCACGAAGCAGGCCAATGGGAGGACATCGATGTCACCTCCAATGACTAG
 GGT

EMX1 antisense

ACCCTAGTCATTGGAGGTGACATCGATGTCCTCCCCATTGGCCTGTTGTGGCAATGCGCCACCG
 GTTGATGTGATGGGAGCCCTTCTCTGCTCAAACACTAGGCCCTCCTCCAGCTCTGCCGT
 TTGTACTTGTCCCTCCGGTTCTGGAACCACACCTCACCTGGCCAGGGAGGGACAGATGA
 A

HEK293 site 3 sense

CATGCAATTAGTCTATTCTGCTGCAAGTAAGCATGCATTGTAGGCTTGATGCTTTCTGCTTCT
 CCAGCCCTGGCCTGGTCAATCCTGGGGCTTAGACTGAGCACGTGATGGCAGAGGAAAGGAAGC
 CCTGCTTCTCCAGAGGGCGTCGCAAGGACAGCTTCTAGACAGGGCTAGTATGTCAGCTCCT

HEK293 site 3 antisense

AGGAGCTGCACATACTAGCCCTGTCTAGGAAAAGCTGCTCGACGCCCTGGAGGAAGCAGG
 GCTTCCTTCCTGCCATACGTGCTCAGTCTAACGCCCCAAGGATTGACCCAGGCCAGGGCTGGA
 GAAGCAGAAAAAAAGCATCAAGCCTACAAATGCATGCTTACTGCAAGCAAATAGACTAATTGCATG

HEK293 site 4 sense

GGCTGACAAAGGCCGGCTGGGTGGAAGGAAGGGAGGAAGGGCGAGGCAGAGGGTCAAAGCAG
 GATGACAGGCAGGGGCACCGCGGCCGGTGGCATTGCGGCTGGAGGTGGGGTTAAAGCGG
 AGACTCTGGTGTGACTACAGTGGGGCCCTGCCCTCTGAGCCCCGCCCTCCAGGCCTGT
 GTGTGT

HEK293 site 4 antisense

ACACACACAGGCCTGGAGGCCGGGCTCAGAGAGGGCAGGGCCCCACTGTAGTCACACAGCACC
 AGAGTCTCCGCTTAAACCCCCACCTCCAGCCGAATGCCACGGGGCGCCGGTGCCCTGCCT
 GTCATCCTGCTTGGACCCCTGCCTGCCCTCCCTCCACCCAGGCCGGCTTGTCA
 GCC

APOE4 sense

AGCACCGAGGAGCTGCGGGTGCCTCGCCTCCCACCTCGCGCAAGCTCGTAAGCGGCTCCTCCG
 CGATGCCGATGACCTGCAGAAGTGCCTGGCAGTGTACCGAGGCCGGCCGAGGGCGCCGAG

CGCGGCCTCAGGCCATCCCGAGCGCCTGGGCCCCCTGGTGGAACAGGGCCGCGTCGGCGCG
CCACTGT

APOE4 antisense

ACAGTGGCGGCCCGCACGCCCTGTTACCAGGGCCCCAGGCCTCGCGGATGGCGCTGA
GGCCGCGCTCGGCCCTCGGGCCCCCTGGTACACTGCCAGGCACCTCTGCAGGTATCG
GCATCGCGAGGAGCCGCTACGCAGCTGCGCAGGTGGAGGCGAGGCACCCGAGCTCCT
CGGTGCT

p53 Y163C sense

ACTCCCCTGCCCTAACAAAGATGTTTGCCAAGACCTGCCCTGTGCAGCTGTGGGTTGA
TTCCACACCCCCGCCGGCACCGCGTCCGCCATGCCATCTACAAGCAGTCACAGCACATGAC
GGAGGTTGTGAGGCCTGCCACCAGTCAGCGCTGCTCAGATAGCGATGGTGAGCAGCTGGGC
TG

p53 Y163C antisense

CAGCCCCAGCTGCTCACCATCGCTATCTGAGCAGCGCTCATGGTGGGGCAGCGCCTCACAAACCTC
CGTCATGTGCTGTGACTGCTGTAGATGCCATGGCGCGACGCCGGTGCAGGGGGGTGTTGG
AATCAACCCACAGCTGCACAGGCAGGTCTGCCAGTTGGCAAAACATCTTGTGAGGGCAGGG
AGT

Deaminase gene gBlocks gene fragments.

hAID

CATCCTTGGTACCGAGCTCGGATCCAGCCACCATGGATAGCCTTGTGAATAGACGCAAGTTCT
GTATCAGTTAAAAACGTGAGATGGCAAAGGCCGACGAGAGACATATCTGTGCTATGCGTTAAG
CGCAGAGATTCCAGGCCACCAAGTTCTCTCGACTCGGCTACCTGCCAACAGAACATGGTTGCCATG
TTGAGCTCCTGTTCTGAGGTATATCAGCGACTGGGATTGGACCCAGGGCGGTGCTATAGGGTGA
CATGGTTACCTCCTGGTCACCTGTTATGACTCGCGCGGCATGTTGCCATTCTGAGAGGGAA
CCCTAACCTGTCTGAGGATCTCACCGCGCAGTGTACTTCTGAGGACCGGAAAGCCGAACC
CGAGGGACTGAGACGCCAACAGAGCGGGTGTGCAGATTGCCATAATGACCTTAAGGACTACT
CTACTGCTGGAACACCTCGTCAAGGACTTCAGGCAGCTCGCCGCATTCTCCGGTGTACGAGGTTGATGACCTCA
GAGATGCCCTTAGAACACTGGACTGTAGGCAGCCGCTCGATTGGTTGGTGTGGCTCAA

rAPOBEC1 (mammalian)

CATCCTTGGTACCGAGCTCGGATCCAGCCACCATGAGCTCAGAGACTGGCCAGTGGCTGTGGACC
CCACATTGAGACGGCGATCGAGCCCCATGAGTTGAGGTATTCTCGATCCGAGAGAGCTCCGCA
AGGAGACCTGCCTGCTTACGAAATTAAATTGGGGGGCCGGCACTCCATTGGCGACATACATCACA
GAACACTAACAGCACGTCGAAGTCACCTCATCGAGAACGTTACGACAGAAAGATATTCTGCG
AACACAAGGTGCAGCATTACCTGGTTCTCAGCTGGAGCCCATGCCGAATGTAGTAGGGCCATC
ACTGAATTCTGTCAAGGTATCCCCACGTCACTCTGTTATTACATCGCAAGGCTGTACCAACACGC
TGACCCCCGCAATCGACAAGGCTGCCGGATTGATCTCTCAGGTGTGACTATCAAATTATGACT
GAGCAGGAGTCAGGATACTGCTGGAGAAACTTGTGAATTATAGCCGAGTAATGAAGCCCAGTGG
CCTAGGTATCCCCATCTGTGGGTACGACTGTACGTTCTGAACTGTACTGCATCATACTGGCCTGC
CTCCTGTCTCAACATTCTGAGAAGGAAGCAGCCACAGCTGACATTCTTACCATCGCTTCAGTCT
TGTCATTACCAGCGACTGCCACACATTCTGGCCACCGGGTTGAAATGAGCGGCCGCTCGA
TTGGTTGGTGTGGCTCAA

pmCDA1

CATCCTTGGTACCGAGCTCGGATCCAGCCACCATGACAGACGCTGAATATGTTAGGATCCATGAAAA
ACTGGATATCTATACATTAAAGAACGAGCTTCTCAATAACAAAAAGTCAGTATCTCACAGATGCTATGT
CCTGTTGCAACTCAAGAGAACGAGGAGAAAGGCCGGCTGTTCTGGGGTACGCCGTTAATAAACC
CCAGTCCGGGACCGAGAGGGGGATTACGCCAGATCTTCAATTAGGAAGGTTGAAGAGTATCT
TCGCGACAATCCCGGTAGTCACAATTACTGGTACAGCTCCTGGAGCCCTGCGCTGATTGCGCC

GAGAAAATACTCGAATGGTACAACCAGGAGTTGAGAGGAATGCCACACTCTCAAGATTGGCTT
GCAAGCTTTACTACGGAGAACGCGAGAAATCAGATTGGCTTGGAACCTCAGGGACAACGGGG
TCGGGTTGAATGTTATGGTGTCCGAACATTACCACTGCTGTAGAAAGATCTTCATTCACTGCAGTCAC
AATCAGCTGAACGAGAACAGATGGCTGGAGAAAACACTGAAACGGGCAGAGAGAAAAGGCAGTCAGAG
CTGAGTATCATGATCCAGGTCAAAATCCTGCATACAACCAAAAGCCGGCTGTATAAGCGGCCGCTC
GATTGGTTGGTGTGGCTCAA

hAPOBEC3G

CATCCTTGGTACCGAGCTGGATCCAGCCACCATGGAGCTGAAGTATCACCTGAGATGCCGGTTTT
CCACTGGTTAGTAAGTGGCGAAACTTCATCGGATCAGGAGTATGAAGTGACCTGGTATATCTCT
TGGTCTCCCTGCACAAAATGTACACCGCAGATGCCACATTCTGGCCGAGGATCCAAGGTGACG
CTCACAATCTTGTGGCCCGCTGTATTATTCTGGACCCGGATTATCAGGAGGCACTTAGGTCT
TGTGCCAAAAGCGCGACGGACCACGGCGACTATGAAAATCATGAATTATGACGAATTCCAGCATTG
CTGGAGTAAGTTGTGTACAGCCAGCGGGAGCTGTTGAGGCCCTGGAACAATCTTCCAAGTACTAC
ATACTGCTTCACATTATGTTGGGGAGATCCTCGGCACTCTATGGATCCTCACCTTACGTTAA
CTTAATAATGAGCCTGGGTCGCGGGCGCCATGAAACCTATTGTGCTACGAGGTCGAGCGGATG
CATATAATGATACGTGGGTCGCTGAATCAGAGGGAGGGGTTCTGTGTAACCAGGCTCCACATAAAC
ATGGATTCTCGAGGGCGGCACGCCAAGTGTGTTCTTGATGTGATACCTTCTGGAAGCTCGA
CCTTGATCAAGATTACAGGGTGACGTGTTCACCTCCTGGTCACCTGCTTCAAGTTGCGCCCAAGAG
ATGGCTAAATTATCAGTAAGAACAAAGCATGTGCCCCCTGTATTTCACAGCCAGAAATTATGATGAC
CAGGGCCGGTGCAGGAGGGCTGCGGACACTCGCTAGGGCGGCAGAAGATCAGCATAATGA
CATACTCCGAATTCAAACACTGTTGGGACACTTTGTGGACCACCAGGGCTGCCATTCAAGCGTG
GGATGGGCTGACGAACATAGTCAGGATCTCTCAGGCCGGCTGCGAGCCATATTGCAGAACCAAGGA
GAATTAGGCGGCCGCTGATTGGTTGGTGTGGCTCAA

rAPOBEC1(E. Coli)

GGCCGGGGATTCTAGAAATAATTTGTTAACCTTAAGAAGGAGATACCATGGATGTCTCTGAAA
CCGGTCCGGTTGCGGTTGACCCGACCCCTCGCTCGTATCGAACCGCACGAATTCAAGTTCT
TCGACCCCGCTGAACTCGCTAAAGAACCTCGCTGTACGAAATCAACTGGGGTGGTCGTCACT
CTATCTGGCGTCACACCTCTCAGAACACCAACAAACACGTTGAAGTTAACCTCATCGAAAATTCA
ACCGAACGTTACTCTGCCAACACCCGTTGCTCTACCTGGTTCTGTCTGGCTCCGTGCG
GTGAATGCTCTCGCGATACCGAATTCTGCTCGTTACCGCACGTTACCGTCAAGGTCTGCGTAC
GCGCGTGTACCAACCACGCGGACCCCGCGTAACCGTCAGGGTCTGCGTACCTGATCTCTGGT
GTTACCATCCAGATCATGACCGAACAGGAATCTGGTACTGCTGGCTAACCTCGTTAACTACTCTCC
GTCTAACGAAGCGCACTGGCCCGTACCCGCACCTGTGGGTTCTGTACGTTCTGGAACGTGA
CTGCATCATCCTGGGTCTGCCCGCTGCCTGAACATCCTCGCTCGTAAACAGCCGAGCTGACCTT
CTTCACCATCGCGCTGCAGTCTGCCACTACCAGCGTCTGCCGCACATCCTGTGGCGACCGG
TCTGAAAGGTGGTAGTGGAGGGAGCGGGCGTTCAATGGATAAGAAATAC

Amino Acid Sequences of BE1, BE2, and BE3.

BE1 for *E. Coli* expression (His₆-rAPOBEC1-XTEN-dCas9)

MGSSHHHHHMSSETGPVAVDPTLRRRIEPHEFEVFFDPRELRKETCLLYEINWGRHSIWRHTSQNTN
KHVEVNFIKEFTTERYFCPNTRCSITWFWSWSPCGECSRAITEFLSRYPHVTLFYIARLYHHADPRNRQGL
RDLISSGVTIQIMTEQESGYCWRNFVNYSRSPSNEAHWPRYPHLWVRLYVLELYCIILGLPPCLNLRRKQPQ
LTFFTIALQSCHYQRLLPHELWATGLKSGSETPGTSESATPESDKKYSIGLAIGTNSVGWAVITDEYKVPSK
KFKVLGNTDRHSIKKNLIGALLFDSGETAETRLKRTARRRYTRRKNRICYLQEIFSNEAKVDDSSFFHRL
EESFLVEEDKKHERHPIFGNIVDEVAYHEKYPTIYHLRKKLVDSTDKAIDLRLIYLALAHMIFRGHFIEGDL
NPNDNSDVDFKLIQLVQTYNQLFEENPINASGVDAKIALSARLSKSRRLENLIAQLPGEKKNGLFGNLI
GLTPNFKSNFDLAEDAKLQLSKDTYDDDNLLAQIGDQYADLFLAAKNLSDAILLSDILRVNTEITKAPLSA
SMIKRYDEHHQDLTLLKALVRQQLPPEKYKEIFFDQSNSKNGYAGYIDGGASQEEFYKFIPILEKMDGTEELL
VKLNREDLLRKQRTFDNGSIPHQIHLGELHAILRRQEDFYPFLKDNRREKIEKILTFRIPYYVGPLARGNSRF
AWMTRKSEETITPWNFEEVVDKGASAQSFIERMTNFDFKLPNEKVLPKHSLLYEYFTVYNELTKVKYVTE
GMRKPAFLSGEQKKAIVDLLFKTNRKVTVKQLKEDYFKKIECFDSVEISGVEDRFNASLGTYHDLLKII
DFLDNEENEDILEDIVLTTLFEDREMIEERLKTYAHLFDDKVMQLKRRRTGWGRSLRKLINGIRDKQS
GKTIIDFLKSDGFANRNFMQLIHDDSLTFKEDIQKAQVSGQQGDSLHEHIANLAGSPAIIKGILQTVKVVDEL

VKVMGRHKPENIVIEMARENQTTQKGQKNSRERMKRIEEGIKELGSQILKEHPVENTQLQNEKLYLYLQNGRDMDYVDQELDINRLSDYDVAPQSFLKDDSIDNKVLTRSDKNRGKSDNVPSEEVKKMKNYWRQLNAKLITQRKFNDLTKAERGGLSELDKAGFIKRQLVETRQITKHVAQILDSRMNTKYDENDKLIREVKVITLKSCLVSDFRKDFQFYKVREINNYHHAHDAYLNAVVGTLAKKYPKLESEFVYGDYKVDVRKMIAKSEQEIGKATAKYFFYSNIMNFFKTEITLANGEIRKRPLIETNGETGEIVWDKGRDFATVRKVL SMPQVNIVKKTEVQTGGFSKESILPKRNSDKLIARKKDWPCKYGGFDSPVTAVSVLVAKVEKGKSKKLKSVKELLGITIMERSSFEKNPIDFLEAKGYKEVKKDLIILPKYSLFELENGRKMLASAGELQKGNEALPSKYVNFLYASHYEKLKGSPEDNEQKQLFVEQHKhYLDEIIEQISEFSKRVILADANLDKVL SAYNKHRDKPIREQAENIIHLFTLTNLGAPAAFKYFDTTIDRKRYTSTKEVLDATLHQHSITGLYETRIDLSQLGGDGGSPKKKRKV

BE1 for Mammalian expression (rAPOBEC1-XTEN-dCas9-NLS)

MSSETGPVAVDPTLRRRIEPHEFEVFFDPRELRKETCLLYEINWGGRHSIWRHTSQNTNKHVEVNIEKF TTERYFCPNTRCSITWFLSWSPCGEC SRAITEFLSRYPHVTLFYIARLYHHADPRNRQGLRDLISSGVTIQ IMTEQESGYCWRNFVNSPSNEAHWPRYPHLWVRLYVLEYCIILGLPPCLNILRRKQPQLTFFTIALQSC HYQRLPPHILWATGLKSGSETPGTSESATPESDKKYSIGLAIGTNSVGWAVITDEYKVPSSKKFKVLGNTDR HSICKNLIGALLFDGETAEATRLKRTARRRYTRRKNRICYLQEIFSNEAKVDDSSFFHRLLEESFLVEEDKK HERHPIFGNIVDEVAYHEKYPTIYHLRKKLVDSTDKA DLRLIYLALAHM IKFRGHFLIEGDLNPNSDVKLF IQLVQTYNQLFEENPINASGVDAKILSARLSKSRRLENLIAQLPGEKKNGLFGNLIALSLGLTPNFKSNFDL AEDAKLQLSKDTYDDDLDNLLAQIGDQYADLFLA AKNLSDAILSDILRVNTEITKAPLSASMIKRYDEHHQ DLTLLKALVRQQLPEKYKEIFFDQS KNGYAGYIDGGASQEEFYKFIKPILEKMDGTEELLVKLNR EDDLLRKQ RTFDNGSIPHQIHLGELHAILRRQEDFYPFLKDNREKIEKILTFRIPYYVGPLARGNSRFAMTRKSEETITP WNFEEVVDKGASAQS FIERMTNFDKNLPNEKVL PKHSLLYEYFTVYNELTKV KYVTEGMRKPAFLSGEQ KKAIVDLLFKTNRKVTVKQLKEDYFKKIECFDSV EISGVEDRFN ASLG TYHDLLKIIKDKDFLDNEENEDILE DIVLTTLFEDREMIEERLKYAHLFDDKVMKQLKRRRYTGWGRLSRKLINGIRDQSGKTILD FLKSDGFA NRNFMQ LIHDDSLTFKEDIQKAQVSGQGDSLHEHIANLAGSPA IKKG ILQTV KVVD ELVK VMGRH K PENIVI EMARENQTTQKGQKNSRERMKRIEEGIKELGSQILKEHPVENTQLQNEKLYLYLQNGRDMDYVDQELDI NRLSDYDVAIVPQSFLKDDSIDNKVLTRSDKNRGKSDNVPSEEVVKKMKNYWRQLLNAKLITQRKF DNLTKAERGGLSELDKAGFIKRQLVETRQITKHVAQILDSRMNTKYDENDKLIREVKVITLKS KLVDFFR KDFQF YKVR EIN NYHH AH DAY LNAV VGT ALI KK YPK LESE FVYGDYKVD VRK MIAK SEQ EIG KATA KYFF YS NIM NFF KTE IT LAN GEIR KRPLIET NG ET GEIVWDKGRDFATVRKVL SMPQVNIVKKTEVQTGGFSKESILPKR NSDKLIARKKDWPCKYGGFDSPVTAVSVLVAKVEKGKSKKLKSVKELLGITIMERSSFEKNPIDFLEAKGYKEVKKDLIILPKYSLFELENGRKMLASAGELQKGNEALPSKYVNFLYASHYEKLKGSPEDNEQKQLFVEQHKhYLDEIIEQISEFSKRVILADANLDKVL SAYNKHRDKPIREQAENIIHLFTLTNLGAPAAFKYFDTTI DRKRYTSTKEVLDATLHQHSITGLYETRIDLSQLGGDGGSPKKKRKV

BE2 (rAPOBEC1-XTEN-dCas9-UGI-NLS)

MSSETGPVAVDPTLRRRIEPHEFEVFFDPRELRKETCLLYEINWGGRHSIWRHTSQNTNKHVEVNIEKF TTERYFCPNTRCSITWFLSWSPCGEC SRAITEFLSRYPHVTLFYIARLYHHADPRNRQGLRDLISSGVTIQ IMTEQESGYCWRNFVNSPSNEAHWPRYPHLWVRLYVLEYCIILGLPPCLNILRRKQPQLTFFTIALQSC HYQRLPPHILWATGLKSGSETPGTSESATPESDKKYSIGLAIGTNSVGWAVITDEYKVPSSKKFKVLGNTDR HSICKNLIGALLFDGETAEATRLKRTARRRYTRRKNRICYLQEIFSNEAKVDDSSFFHRLLEESFLVEEDKK HERHPIFGNIVDEVAYHEKYPTIYHLRKKLVDSTDKA DLRLIYLALAHM IKFRGHFLIEGDLNPNSDVKLF IQLVQTYNQLFEENPINASGVDAKILSARLSKSRRLENLIAQLPGEKKNGLFGNLIALSLGLTPNFKSNFDL AEDAKLQLSKDTYDDDLDNLLAQIGDQYADLFLA AKNLSDAILSDILRVNTEITKAPLSASMIKRYDEHHQ DLTLLKALVRQQLPEKYKEIFFDQS KNGYAGYIDGGASQEEFYKFIKPILEKMDGTEELLVKLNR EDDLLRKQ RTFDNGSIPHQIHLGELHAILRRQEDFYPFLKDNREKIEKILTFRIPYYVGPLARGNSRFAMTRKSEETITP WNFEEVVDKGASAQS FIERMTNFDKNLPNEKVL PKHSLLYEYFTVYNELTKV KYVTEGMRKPAFLSGEQ KKAIVDLLFKTNRKVTVKQLKEDYFKKIECFDSV EISGVEDRFN ASLG TYHDLLKIIKDKDFLDNEENEDILE DIVLTTLFEDREMIEERLKYAHLFDDKVMKQLKRRRYTGWGRLSRKLINGIRDQSGKTILD FLKSDGFA NRNFMQ LIHDDSLTFKEDIQKAQVSGQGDSLHEHIANLAGSPA IKKG ILQTV KVVD ELVK VMGRH K PENIVI EMARENQTTQKGQKNSRERMKRIEEGIKELGSQILKEHPVENTQLQNEKLYLYLQNGRDMDYVDQELDI NRLSDYDVAIVPQSFLKDDSIDNKVLTRSDKNRGKSDNVPSEEVVKKMKNYWRQLLNAKLITQRKF DNLTKAERGGLSELDKAGFIKRQLVETRQITKHVAQILDSRMNTKYDENDKLIREVKVITLKS KLVDFFR KDFQF YKVR EIN NYHH AH DAY LNAV VGT ALI KK YPK LESE FVYGDYKVD VRK MIAK SEQ EIG KATA KYFF YS NIM NFF KTE IT LAN GEIR KRPLIET NG ET GEIVWDKGRDFATVRKVL SMPQVNIVKKTEVQTGGFSKESILPKR NSDKLIARKKDWPCKYGGFDSPVTAVSVLVAKVEKGKSKKLKSVKELLGITIMERSSFEKNPIDFLEAKGYKEVKKDLIILPKYSLFELENGRKMLASAGELQKGNEALPSKYVNFLYASHYEKLKGSPEDNEQKQLFVEQHKhYLDEIIEQISEFSKRVILADANLDKVL SAYNKHRDKPIREQAENIIHLFTLTNLGAPAAFKYFDTTI DRKRYTSTKEVLDATLHQHSITGLYETRIDLSQLGGDGGSPKKKRKV

YKEVKKDIIKLPKYSLEFELENGRKMLASAGELQKGNELALPSKYVNFLYLA SHYEKLKGSPEDNEQKQL
FVEQHKHYLDEIIEQISEFSKRVILADANLDKVLSAYNKHRDKPIREQAENIIHLFTLTNLGAPAAFKYFDTTI
DRKRYTSTKEVLDATLIHQSITGLYETRIDSQLGGDSGGSTNLSDIIEKETGKQLVIQESILMLPEEEV
NKPESDILVHTAYDESTDENVMLLTSDAPEYKPWALVIQDSNGENKIKMLSGGSPKKRKV

BE3 (rAPOBEC1-XTEN-Cas9n-UGI-NLS)

MSSETGPVAVDPTLRRRIEPHEFEVFFDPRELRKETCLLYEINWGGRHSIWRHTSQNTNKHVEVNIEKF
TTERYFCPNTRCSITWFLSWSPCGECSCRAITEFLSRYPHTLFIYIARLYHHADPRNRQGLRDLISSGVTIQ
IMTEQESGYCWRNFVNYSPSNEAHWPRYPHLWVRLYVLELYCIILGPLPCLNIRRKQPQLTFFTIALQSC
HYQRLLPPHILWATGLKSGSETPGTSESATPESDKKYSIGLAIGTNSVGWAVITDEYKVPSKKFVLGNTDR
HSIKKNLIGALLFDGETAEATRLKRTARRRYTRRKNRICYLQEIFSNEMAKVDDSSFFHRLEESFLVEEDKK
HERHPIFGNIVDEVAYHEKYPTIYHLRKKLVSTDKA DLRLIYLALAHMIKFRGHFLIEGDLNPDNSVDKLF
IQLVQTYNQLFEENPINASGVDAKAILSRSRLENLIAQLPGEKKNGLFGNLIALSLGLTPNFKSNFDL
AEDA KLQLSKDTYDDDLDNLLAQIGDQYADLFLA AKNLSDAILLS DILRVNTEITKAPLSASMIKRYDEHHQ
DLTLLKALVRQQQLPEKYKEIFFDQS KNGYAGYIDGGASQEEFYKFIPILEKMDGTEELLV KLNREDLLRKQ
RTFDNGSIPHQIHLGELHAILRRQEDFYPFLKDNREKIEKILTFRIPYYVGPLARGNSRFAMTRKSEETITP
WNFEEVVVDKGASAQS FIERMTNFDKNLPNEKVLPKHSLLYEYFTVYNELTKV KVYTEGMRKPAFLSGEQ
KKAIVDLLFKTNRKVTVKQLKEDYFKKIECFDSVEISGVEDRFN ASL GTYHDLLKIIKDKDFLDNEENEDILE
DIVLTTLFEDREMIEERLKTYAHLFDDKVMKQLKRRRTGWGRLSRKLINGIRDQSGKTILDFLKSDGFA
NRNFMQLIHDDSLTFKEDIQKAQVSGQGDSLHEHIANLAGSPA IKKG ILQTVK VDELVK VMGRHK PENIVI
EMARENQTTQKGQKNSRERMKRIEGIKE LGSQLKEHPVENTQLQNEKLYLYLQNGRD MYVDQELDI
NRLSDYDVHDIVPQSFLKDDSIDNKVLTRSDKNRGKSDNVPSEEVVKMKNYWRQLLNAKLITQRKFDNL
TKAERGGLSELDKAGFIKRQLVETRQITKHVAQILD SRMNTKYDENDKLIREVKVITLKS KLVSDFRKDFQF
YKVREINNYHHAHDAYLNAVVG TALIKKYPKLESEFVYGDYKVYDVRKMIAKSEQEIGKATAKYFFYSNIM
NFFKTEITLANGEIRKRPLIETNGETGEIVWDKGRDFATVRKVL SMPQVNIVKKTEVQTGGFSKESILPKRN
SDKLIARKKDWDPKYGGFDPTVAYS VLVAKVEKGKSKKLKSVKELLGITIMERSSFEKNPIDFLEAKG
YKEVKKDIIKLPKYSLEFELENGRKMLASAGELQKGNELALPSKYVNFLYLA SHYEKLKGSPEDNEQKQL
FVEQHKHYLDEIIEQISEFSKRVILADANLDKVLSAYNKHRDKPIREQAENIIHLFTLTNLGAPAAFKYFDTTI
DRKRYTSTKEVLDATLIHQSITGLYETRIDSQLGGDSGGSTNLSDIIEKETGKQLVIQESILMLPEEEV
NKPESDILVHTAYDESTDENVMLLTSDAPEYKPWALVIQDSNGENKIKMLSGGSPKKRKV

SUPPLEMENTARY TABLES

Supplementary Table 1. Activities of BE1, BE2, and BE3 at EMX1 off-targets. HEK293T cells were transfected with plasmids expressing BE1, BE2, or BE3 and a sgRNA matching the EMX1 sequence using Lipofectamine 2000. Three days after transfection, genomic DNA was extracted, amplified by PCR, and analyzed by high-throughput DNA sequencing at the on-target loci, plus the top ten known Cas9 off-target loci for the EMX1 sgRNA, as previously determined by Joung and coworkers using the GUIDE-seq method (main text Ref. 19). EMX1 off-target 5 locus did not amplify and is not shown. Sequences of the on-target and off-target protospacers and protospacer adjacent motifs (PAMs) are displayed. Cellular C to T conversion percentages, defined as the percentage of total DNA sequencing reads with T at each position of an original C within the protospacer, are shown for BE1, BE2, and BE3. On the far right are displayed the total number of sequencing reads reported by Joung and coworkers for each sequence.

Supplementary Table 2. Activities of BE1, BE2, and BE3 at FANCF off-targets. HEK293T cells were transfected with plasmids expressing BE1, BE2, or BE3 and a sgRNA matching the FANCF sequence using Lipofectamine 2000. Three days after transfection, genomic DNA was extracted, amplified by PCR, and analyzed by high-throughput DNA sequencing at the on-target loci, plus all of the known Cas9 off-target loci for the FANCF sgRNA, as previously determined by Joung and coworkers using the GUIDE-seq method (main text Ref. 19). Sequences of the on-target and off-target protospacers and protospacer adjacent motifs (PAMs) are displayed. Cellular C to T conversion percentages, defined as the percentage of total DNA sequencing reads with T at each position of an original C within the protospacer, are shown for BE1, BE2, and BE3. On the far right are displayed the total number of sequencing reads reported by Joung and coworkers for each sequence.

	GUIDE-seq counts																																											
FANCF on target	<table border="1"><tr><td>G</td><td>G</td><td>A</td><td>A</td><td>T</td><td>C</td><td>C</td><td>C</td><td>T</td><td>T</td><td>C</td><td>T</td><td>G</td><td>C</td><td>A</td><td>G</td><td>C</td><td>A</td><td>C</td><td>C</td><td>T</td><td>G</td><td>G</td></tr></table>																				G	G	A	A	T	C	C	C	T	T	C	T	G	C	A	G	C	A	C	C	T	G	G	
G	G	A	A	T	C	C	C	T	T	C	T	G	C	A	G	C	A	C	C	T	G	G																						
untreated	<table border="1"><tr><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td></tr></table>																				0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0			
0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0																								
BE1	<table border="1"><tr><td>3.7</td><td>3.2</td><td>3.4</td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td>2.4</td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td></tr></table>																				3.7	3.2	3.4								2.4													
3.7	3.2	3.4								2.4																																		
BE2	<table border="1"><tr><td>4.1</td><td>3.5</td><td>3.5</td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td>0.4</td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td></tr></table>																				4.1	3.5	3.5								0.4													
4.1	3.5	3.5								0.4																																		
BE3	<table border="1"><tr><td>19.1</td><td>16.0</td><td>16.7</td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td>8.8</td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td></tr></table>																				19.1	16.0	16.7								8.8													
19.1	16.0	16.7								8.8																																		
FANCF off target 1	<table border="1"><tr><td>G</td><td>G</td><td>A</td><td>A</td><td>C</td><td>C</td><td>C</td><td>C</td><td>G</td><td>T</td><td>C</td><td>T</td><td>G</td><td>C</td><td>A</td><td>G</td><td>C</td><td>A</td><td>C</td><td>C</td><td>A</td><td>G</td><td>G</td></tr></table>																				G	G	A	A	C	C	C	C	G	T	C	T	G	C	A	G	C	A	C	C	A	G	G	2099
G	G	A	A	C	C	C	C	G	T	C	T	G	C	A	G	C	A	C	C	A	G	G																						
untreated	<table border="1"><tr><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td></tr></table>																				0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0			
0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0																								
BE1	<table border="1"><tr><td>0.0</td><td>0.4</td><td>0.0</td><td>0.1</td><td></td><td></td><td></td><td></td><td></td><td></td><td>0.0</td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td></tr></table>																				0.0	0.4	0.0	0.1							0.0													
0.0	0.4	0.0	0.1							0.0																																		
BE2	<table border="1"><tr><td>0.2</td><td>0.2</td><td>0.3</td><td>0.3</td><td></td><td></td><td></td><td></td><td></td><td></td><td>0.0</td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td></tr></table>																				0.2	0.2	0.3	0.3							0.0													
0.2	0.2	0.3	0.3							0.0																																		
BE3	<table border="1"><tr><td>1.9</td><td>2.1</td><td>1.5</td><td>1.3</td><td></td><td></td><td></td><td></td><td></td><td></td><td>1.3</td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td></tr></table>																				1.9	2.1	1.5	1.3							1.3													
1.9	2.1	1.5	1.3							1.3																																		
FANCF off target 2	<table border="1"><tr><td>G</td><td>G</td><td>A</td><td>G</td><td>T</td><td>C</td><td>C</td><td>C</td><td>T</td><td>C</td><td>C</td><td>T</td><td>A</td><td>C</td><td>A</td><td>G</td><td>C</td><td>A</td><td>C</td><td>C</td><td>A</td><td>G</td><td>G</td></tr></table>																				G	G	A	G	T	C	C	C	T	C	C	T	A	C	A	G	C	A	C	C	A	G	G	524
G	G	A	G	T	C	C	C	T	C	C	T	A	C	A	G	C	A	C	C	A	G	G																						
untreated	<table border="1"><tr><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td></tr></table>																				0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0			
0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0																								
BE1	<table border="1"><tr><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td></tr></table>																				0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0			
0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0																								
BE2	<table border="1"><tr><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td></tr></table>																				0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1			
0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1																								
BE3	<table border="1"><tr><td>0.2</td><td>0.2</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td><td>0.1</td></tr></table>																				0.2	0.2	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1			
0.2	0.2	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1	0.1																								
FANCF off target 3	<table border="1"><tr><td>A</td><td>G</td><td>A</td><td>G</td><td>G</td><td>C</td><td>C</td><td>C</td><td>C</td><td>T</td><td>C</td><td>T</td><td>G</td><td>C</td><td>A</td><td>G</td><td>C</td><td>A</td><td>C</td><td>C</td><td>A</td><td>G</td><td>G</td></tr></table>																				A	G	A	G	G	C	C	C	C	T	C	T	G	C	A	G	C	A	C	C	A	G	G	150
A	G	A	G	G	C	C	C	C	T	C	T	G	C	A	G	C	A	C	C	A	G	G																						
untreated	<table border="1"><tr><td>0.0</td><td>0.1</td><td>0.1</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td></tr></table>																				0.0	0.1	0.1	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0			
0.0	0.1	0.1	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0																								
BE1	<table border="1"><tr><td>0.0</td><td>0.1</td><td>0.1</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td></tr></table>																				0.0	0.1	0.1	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0			
0.0	0.1	0.1	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0																								
BE2	<table border="1"><tr><td>0.0</td><td>0.1</td><td>0.1</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.1</td><td>0.0</td><td>0.0</td></tr></table>																				0.0	0.1	0.1	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.1	0.0	0.0			
0.0	0.1	0.1	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.1	0.0	0.0																								
BE3	<table border="1"><tr><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td></tr></table>																				0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0			
0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0																								
FANCF off target 4	<table border="1"><tr><td>A</td><td>C</td><td>C</td><td>A</td><td>T</td><td>C</td><td>C</td><td>C</td><td>T</td><td>C</td><td>C</td><td>T</td><td>G</td><td>C</td><td>A</td><td>G</td><td>C</td><td>A</td><td>C</td><td>C</td><td>A</td><td>G</td><td>G</td></tr></table>																				A	C	C	A	T	C	C	C	T	C	C	T	G	C	A	G	C	A	C	C	A	G	G	125
A	C	C	A	T	C	C	C	T	C	C	T	G	C	A	G	C	A	C	C	A	G	G																						
untreated	<table border="1"><tr><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td></tr></table>																				0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0			
0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0																								
BE1	<table border="1"><tr><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.2</td><td>0.0</td></tr></table>																				0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.2	0.0			
0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.2	0.0																								
BE2	<table border="1"><tr><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td></tr></table>																				0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0			
0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0																								
BE3	<table border="1"><tr><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td></tr></table>																				0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0			
0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0																								
FANCF off target 5	<table border="1"><tr><td>G</td><td>G</td><td>A</td><td>T</td><td>T</td><td>G</td><td>C</td><td>C</td><td>A</td><td>T</td><td>C</td><td>C</td><td>G</td><td>C</td><td>A</td><td>G</td><td>C</td><td>A</td><td>C</td><td>C</td><td>T</td><td>G</td><td>G</td></tr></table>																				G	G	A	T	T	G	C	C	A	T	C	C	G	C	A	G	C	A	C	C	T	G	G	101
G	G	A	T	T	G	C	C	A	T	C	C	G	C	A	G	C	A	C	C	T	G	G																						
untreated	<table border="1"><tr><td>0.1</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td></tr></table>																				0.1	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0			
0.1	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0																								
BE1	<table border="1"><tr><td>0.1</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td></tr></table>																				0.1	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0			
0.1	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0																								
BE2	<table border="1"><tr><td>0.1</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0.0</td></tr></table>																				0.1	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0			
0.1	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0																								
BE3	<table border="1"><tr><td>0.1</td><td>0.0</td><td>0.0</td><td>0.0</td><td>0</td></tr></table>	0.1	0.0	0.0	0.0	0																																						
0.1	0.0	0.0	0.0	0																																								

Supplementary Table 3. Activities of BE1, BE2, and BE3 at HEK293 site 2 off-targets. HEK293T cells were transfected with plasmids expressing BE1, BE2, or BE3 and a sgRNA matching the HEK293 site 2 sequence using Lipofectamine 2000. Three days after transfection, genomic DNA was extracted, amplified by PCR, and analyzed by high-throughput DNA sequencing at the on-target loci, plus all of the known Cas9 and dCas9 off-target loci for the HEK293 site 2 sgRNA, as previously determined by Joung and coworkers using the GUIDE-seq method (main text Ref. 21), and Adli and coworkers using chromatin immunoprecipitation high-throughput sequencing (ChIP-seq) experiments (main text Ref. 31). Sequences of the on-target and off-target protospacers and protospacer adjacent motifs (PAMs) are displayed. Cellular C to T conversion percentages, defined as the percentage of total DNA sequencing reads with T at each position of an original C within the protospacer, are shown for BE1, BE2, and BE3. On the far right are displayed the total number of sequencing reads reported by Joung and coworkers, and the ChIP-seq signal intensity reported by Adli and coworkers for each sequence.

Supplementary Table 4. Activities of BE1, BE2, and BE3 at HEK293 site 3 off-targets. HEK293T cells were transfected with plasmids expressing BE1, BE2, or BE3 and a sgRNA matching the HEK293 site 3 sequence using Lipofectamine 2000. Three days after transfection, genomic DNA was extracted, amplified by PCR, and analyzed by high-throughput DNA sequencing at the on-target loci, plus all of the known Cas9 off-target loci and the top five known dCas9 off-target loci for the HEK293 site 3 sgRNA, as previously determined by Joung and coworkers using the GUIDE-seq method (main text Ref. 21), and Adli and coworkers using chromatin immunoprecipitation high-throughput sequencing (ChIP-seq) experiments (main text Ref. 31). Sequences of the on-target and off-target protospacers and protospacer adjacent motifs (PAMs) are displayed. Cellular C to T conversion percentages, defined as the percentage of total DNA sequencing reads with T at each position of an original C within the protospacer, are shown for BE1, BE2, and BE3. On the far right are displayed the total number of sequencing reads reported by Joung and coworkers, and the ChIP-seq signal intensity reported by Adli and coworkers for each sequence.

GUIDE-seq
counts/ ChIP-seq
intensity
2074/ 163

Supplementary Table 5. Activities of BE1, BE2, and BE3 at HEK293 site 4 off-targets. HEK293T cells were transfected with plasmids expressing BE1, BE2, or BE3 and a sgRNA matching the HEK293 site 4 sequence using Lipofectamine 2000. Three days after transfection, genomic DNA was extracted, amplified by PCR, and analyzed by high-throughput DNA sequencing at the on-target loci, plus the top ten known Cas9 off-target loci and the top five known dCas9 off-target loci for the HEK293 site 4 sgRNA, as previously determined by Joung and coworkers using the GUIDE-seq method (main text Ref. 21), and Adli and coworkers using chromatin immunoprecipitation high-throughput sequencing (ChIP-seq) experiments (main text Ref. 31). Sequences of the on-target and off-target protospacers and protospacer adjacent motifs (PAMs) are displayed. Cellular C to T conversion percentages, defined as the percentage of total DNA sequencing reads with T at each position of an original C within the protospacer, are shown for BE1, BE2, and BE3. On the far right are displayed the total number of sequencing reads reported by Joung and coworkers, and the ChIP-seq signal intensity reported by Adli and coworkers for each sequence.

HEK site 4 ChIP-seq off target 1	<table border="1"><tr><td>G</td><td>T</td><td>G</td><td>G</td><td>C</td><td>T</td><td>G</td><td>G</td><td>A</td><td>G</td><td>G</td><td>T</td><td>G</td><td>G</td><td>A</td><td>G</td><td>G</td><td>T</td><td>G</td><td>G</td><td>G</td><td>G</td><td>G</td></tr></table>	G	T	G	G	C	T	G	G	A	G	G	T	G	G	A	G	G	T	G	G	G	G	G	110
G	T	G	G	C	T	G	G	A	G	G	T	G	G	A	G	G	T	G	G	G	G	G			
untreated	0.0																								
BE1	0.0																								
BE2	0.0																								
BE3	0.0																								
HEK site 4 ChIP-seq off target 3	<table border="1"><tr><td>G</td><td>A</td><td>G</td><td>G</td><td>G</td><td>A</td><td>A</td><td>G</td><td>G</td><td>G</td><td>C</td><td>T</td><td>G</td><td>G</td><td>A</td><td>G</td><td>G</td><td>T</td><td>G</td><td>G</td><td>A</td><td>G</td><td>G</td></tr></table>	G	A	G	G	G	A	A	G	G	G	C	T	G	G	A	G	G	T	G	G	A	G	G	89
G	A	G	G	G	A	A	G	G	G	C	T	G	G	A	G	G	T	G	G	A	G	G			
untreated	0.0																								
BE1	0.0																								
BE2	0.0																								
BE3	0.0																								

Supplementary Table 6: Mutation rates of non-protospacer bases following BE3-mediated correction of the Alzheimer's disease-associated *APOE4* allele to *APOE3r* in mouse astrocytes.

The DNA sequence of the 50 bases on either side of the protospacer from Fig. 4a and Extended Data Fig. 7a is shown with each base's position relative to the protospacer. The side of the protospacer distal to the PAM is designated with positive numbers, while the side that includes the PAM is designated with negative numbers, with the PAM shown in blue. Underneath each sequence are the percentages of total DNA sequencing reads with the corresponding base for untreated cells, for cells treated with BE3 and an sgRNA targeting the *APOE4* C158R mutation, or for cells treated with BE3 and an sgRNA targeting the *VEGFA* locus. Neither BE3-treated sample resulted in mutation rates above those of untreated controls.

<i>APOE4</i>	C50	C49	C48	C47	T46	G45	C44	G43	C42	A41	A40	G39	C38	T37	G36	C35	G34	T33	A32	A31	G30	C29	G28	G27	C26	
Untreated																										
A	0.1±0.1	100±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	
C	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	
G	0.1±0.1	0.1±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.99±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	
T	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	
BE3 + on-target sgRNA																										
A	0±0	100±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1
C	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0±0	0.1±0.1	100±0.1	0±0	0.1±0.1	100±0.1	0±0
G	0.1±0.1	0.1±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1
T	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1
BE3 + off-target sgRNA																										
A	0±0	100±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1
C	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1
G	0±0	0.1±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0±0	0.1±0.1	100±0.1	0±0	0.1±0.1	100±0.1	0±0
T	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1
<i>APOE4</i>	T25	C24	C23	T22	C21	C20	G19	C18	G17	A16	T15	G14	C13	C12	G11	A10	T9	G8	A7	C6	T4	G3	C2	A1		
Untreated																										
A	0.1±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1
C	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1
G	0±0	0.1±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0±0	0.1±0.1	100±0.1	0±0	0.1±0.1	100±0.1	0±0
T	100±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1
BE3 + on-target sgRNA																										
A	0±0	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1
C	0.1±0.1	99.9±0.1	1.100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1
G	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1
T	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1
BE3 + off-target sgRNA																										
A	0.1±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0±0	0.1±0.1	100±0.1	0±0	0.1±0.1	100±0.1	0±0
C	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1
G	0±0	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1
T	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1
<i>APOE4</i>	A-1	G-2	G-3	C-4	C-5	G-6	G-7	G-8	G-9	C-10	C-11	C-12	G-13	C-14	G-15	A-16	G-17	G-18	G-19	C-20	G-21	C-22	C-23	G-24	A-25	
Untreated																										
A	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1
C	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1
G	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1	0.1±0.1	100±0.1	0.1±0.1

Supplementary Table 7: Mutation rates of non-protospacer bases following BE3-mediated correction of the cancer-associated p53 Y163C mutation in HCC1954 human cells. The DNA sequence of the 50 bases on either side of the protospacer from Fig. 4b and Extended Data Fig. 7b is shown with each base's position relative to the protospacer. The side of the protospacer distal to the PAM is designated with positive numbers, while the side that includes the PAM is designated with negative numbers, with the PAM shown in blue. Underneath each sequence are the percentages of total sequencing reads with the corresponding base for untreated cells, for cells treated with BE3 and an sgRNA targeting the TP53 Y163C mutation, or for cells treated with BE3 and an sgRNA targeting the VEGFA locus. Neither BE3-treated sample resulted in mutational rates above those of untreated controls.

Supplementary Table 8: List of 911 base-editable gene variants associated with human disease with an NGG PAM positioned appropriately. Shown for each gene variant are, from left to right, the Single Nucleotide Polymorphism Database (dbSNP) identification number, the genotype (written as the NCBI GenBank identification number of the gene, the gene name, the chromosome location and DNA base substitution of the SNP, and the amino acid substitution caused by the SNP), the protospacer and PAM sequence(s) that would be used in combination with a base editor to correct each SNP (shown as the coding strand sequence), and the associated genetic disease. The activity window was defined as positions 4-8. Variants containing only a single C within the activity window are highlighted in yellow (284 variants). We note that Cas9 variants with altered PAM specificities may enable many disease-associated gene variants not listed below to be addressed by base editing.

dbSNP #	Genotype	Protospacer and PAM sequence(s)	Associated genetic disease
755445790	NM_000391.3(TPP1):c.887-10A>G	TTTYTTTTTTTTTTTTTGAGG	Ceroid lipofuscinosis, neuronal, 2
113994167	NM_000018.3(ACADVL):c.848T>C (p.Val283Ala)	TTTGGGTGGAGAGGGGCTTCGG, TTGGTGGAGAGGGGCTTCGGG	Very long chain acyl-CoA dehydrogenase deficiency
119470018	NM_024996.5(GFM1):c.521A>G (p.Asn174Ser)	TTGYTAATAAAAGTTAGAACGG	Combined oxidative phosphorylation deficiency 1
115650537	NM_000426.3(LAMA2):c.8282T>C (p.Ile2761Thr)	TTGAYAGGGAGCAAGCAGTCGG, TGAYAGGGAGCAAGCAGTCGGG	Merosin deficient congenital muscular dystrophy
587777752	NM_014946.3(SPAST):c.1688-2A>G	TTCYGTAAAACATAAAAGTCAGG	Spastic paraparesis 4, autosomal dominant
794726821	NM_001165963.1(SCN1A):c.4055T>C (p.Leu1352Pro)	TTCYGGTTGTCTTATATTCTGG	Severe myoclonic epilepsy in infancy
397514745	NM_001130089.1(KARS):c.517T>C (p.Tyr173His)	CTTCYATGATCTCGAGGAGAGG, TTCYATGATCTCGAGGAGAGGG	Deafness, autosomal recessive 89
376960358	NM_001202.3(BMP4):c.362A>G (p.His121Arg)	TTCGTGGYGGAAGCTCCTCACGG	Microphthalmia syndromic 6
606231280	NM_001287223.1(SCN11A):c.1142T>C (p.Ile381Thr)	CTTCAYTGTGGTCATTTCTGG, TTCAYTGTGGTCATTTCTGGG	Episodic pain syndrome, familial, 3
387906735	m.608A>G	TTCAGYGTATTGCTTGAGGAGG	
199474663	m.3260A>G	TTAACGTTATCGCATTACCGGG	Cardiomyopathy with or without skeletal myopathy
104894962	NM_003413.3(ZIC3):c.1213A>G (p.Lys405Glu)	TGTGTTYGCGCAGGGAGCTCGGG, ATGTGTTYGCGCAGGGAGCTCGG	Heterotaxy, visceral, X-linked
796053181	NM_021007.2(SCN2A):c.1271T>C (p.Val424Ala)	TGTGGYGGCCATGCCCTATGAGG	not provided
267606788	NM_000129.3(F13A1):c.728T>C (p.Met243Thr)	TGTGAYGGACAGAGCACAAATGG	Factor xiii, a subunit, deficiency of
397514503	NM_003863.3(DPM2):c.68A>G (p.Tyr23Cys)	TGTAGYAGGTGAAGATGATCAGG	Congenital disorder of glycosylation type 1u
104893973	NM_000416.2(IFNGR1):c.260T>C (p.Ile87Thr)	TGTAATAYTTCTGATCATGTTGG	Disseminated atypical mycobacterial infection, Mycobacterium tuberculosis, susceptibility to
121908466	NM_005682.6(ADGRG1):c.263A>G (p.Tyr88Cys)	TGGYAGAGGCCCTGGGTCAGG	Polymicrogyria, bilateral frontoparietal
147952488	NM_002437.4(MPV17):c.186+2T>C	TGGYAAGTTCTCCCCACAGGG	Navajo neurohepatopathy
121909537	NM_001145.4(ANG):c.121A>G (p.Lys41Glu)	TGGTTYGGCATCATAGTGCCTGGG, GTGTTYGGCATCATAGTGCCTGG	Amyotrophic lateral sclerosis type 9
121918489	NM_000141.4(FGFR2):c.1018T>C (p.Tyr340His)	TGGGGAAAYATACGTGCTTGGCGGG, GGGGAAAYATACGTGCTTGGCGGG	Crouzon syndrome
121434463	m.12320A>G	GACTYGCACCAAAATTGGGG, GGACTYGCACCAAAATTGGGG, TGGAGTYGCACCAAAATTGGGG	Mitochondrial myopathy
121908046	NM_000403.3(GALE):c.101A>G (p.Asn34Ser)	TGGAAGYTATCGATGACCACAGG	UDPGlucose-4-epimerase deficiency
431905512	NM_003764.3(STX11):c.173T>C (p.Leu58Pro)	TGCYGGTGGCCGACGTGAAGCGG	Hemophagocytic lymphohistiocytosis, familial, 4
121917905	NM_000124.3(ERCC6):c.2960T>C (p.Leu987Pro)	TGCYAAAGACCCAAAACAAAGG	Cerebro-oculo-facio-skeletal syndrome
121918500	NM_000141.4(FGFR2):c.874A>G (p.Lys292Glu)	TGCTYGATCCACTGGATGTGGGG, TGCTYGATCCACTGGATGTGGGG, CGTGCTYGATCCACTGGATGTGG	Crouzon syndrome
60431989	NM_000053.3(ATP7B):c.3443T>C (p.Ile1148Thr)	TGCTGAYTGGAAACCGTGAGTGG	Wilson disease
78950939	NM_000250.1(MPO):c.518A>G (p.Tyr173Cys)	GTGCGGGYATTTGCTCTGCTCCGG, TGCAGGYATTTGCTCTGCTCCGGG	Myeloperoxidase deficiency
115677373	NM_201631.3(TGM5):c.763T>C (p.Trp255Arg)	TGCAGGAGYGGACGGCAGCGTG	Peeling skin syndrome, acral type
5030804	NM_000551.3(VHL):c.233A>G (p.Asn78Ser)	GCGAYTGCAGAAGATGACCTGGG, TGCAGTGCAGAAGATGACCTGG	Von Hippel-Lindau syndrome
397508328	NM_000492.3(CFTR):c.1A>G (p.Met1Val)	GCAYGGTCTCTCGGGCGCTGGGG, TGCAYGGTCTCTCGGGCGCTGGG	Cystic fibrosis

		CTGCAYGGTCTCTCGGGCGCTGG	
137853299	NM_000362.4(TIMP3):c.572A>G (p.Tyr191Cys)	TGCAGYAGCCGCCCTCTGCCGG	Sorsby fundus dystrophy
121908549	NM_000334.4(SCN4A):c.3478A>G (p.Ile1160Val)	TGAYGGAGGGATGGCGCCTAGG	
121909337	NM_001451.2(FOXF1):c.1138T>C (p.Ter380Arg)	TGATGYGAGGCTGCCGCCGAGG	Alveolar capillary dysplasia with misalignment of pulmonary veins
281875320	NM_005359.5(SMAD4):c.1500A>G (p.Ile500Met)	TGAGYATGCATAAGCGACGAAGG	Myhre syndrome
730880132	NM_170707.3(LMNA):c.710T>C (p.Phe237Ser)	TGAGTYTGAGAGCCGGCTGGCGG	Primary dilated cardiomyopathy
281875322	NM_005359.5(SMAD4):c.1498A>G (p.Ile500Val)	TGAGTAYGCATAAGCGACGAAGG	Hereditary cancer-predisposing syndrome, Myhre syndrome
72556283	NM_000531.5(OTC):c.527A>G (p.Tyr176Cys)	TGAGGYAATCAGCCAGGATCTGG	not provided
74315311	NM_020435.3(GJC2):c.857T>C (p.Met286Thr)	TGAGAYGGCCCACCTGGGCTTGG, GAGAYGGCCCACCTGGGCTTGGG	Leukodystrophy, hypomyelinating, 2
121912495	NM_170707.3(LMNA):c.1139T>C (p.Leu380Ser)	TCTYGGAGGGCGAGGAGGAGGG	Congenital muscular dystrophy, LMNA-related
128620184	NM_000061.2(BTK):c.1288A>G (p.Lys430Glu)	TCTYGATGGCCACGTCGTACTGG	X-linked agammaglobulinemia
118192252	NM_004519.3(KCNQ3):c.1403A>G (p.Asn468Ser)	TCTTAYTGTAAAGCCAACAGG	Benign familial neonatal seizures 2, not specified
121909142	NM_001300.5(KLF6):c.190T>C (p.Trp64Arg)	TCTGYGGACCAAAATCATTCTGG	
104895503	NM_001127255.1(NLRP7):c.2738A>G (p.Asn913Ser)	TCTGGYTGATACTCAAGTCCAGG	Hydatidiform mole
587783035	NM_000038.5(APC):c.1744-2A>G	TCCYAGTAAGAACAGAACATGG	Familial adenomatous polyposis 1
72556289	NM_000531.5(OTC):c.541-2A>G	TCCYAAAAGGCACGGGATGAAGG	not provided
28937313	NM_005502.3(ABCA1):c.2804A>G (p.Asn935Ser)	TCCAYTGTGGCCAGGAAGGAGG, CGCTCCAYTGTGGCCAGGAAGG	Tangier disease
143246552	NM_001003811.1(TEX11):c.511A>G (p.Met171Val)	TCCAYGGTCAAGTCAGCCTCAGG, CCAYGGTCAAGTCAGCCTCAGGG	Spermatogenic failure, X-linked, 2
587776451	NM_002049.3(GATA1):c.2T>C (p.Met1Thr)	CTCCAYGGAGTCCCTGGCCTGG, TCCAYGGAGTTCCCTGGCCTGGG, CCAYGGAGTTCCCTGGCCTGGGG	GATA-1-related thrombocytopenia with dyserythropoiesis
121908403	NM_021102.3(SPINT2):c.488A>G (p.Tyr163Cys)	TCCAYAGATGAAGTTATTGCAGG	Diarrhea 3, secretory sodium, congenital, syndromic
281874738	NM_000495.4(COL4A5):c.438+2T>C	CTCCAGYAAGTTAAAAATTGG, TCCAGYAAGTTAAAAATTGGG	Alport syndrome, X-linked recessive
730880279	NM_030653.3(DDX11):c.2271+2T>C	TCCAGGYGCGGGCGTCATGCTGG, CCAGGYGGCGGGCGTCATGCTGGG	Warsaw breakage syndrome
28940272	NM_017890.4(VPS13B):c.8978A>G (p.Asn2993Ser)	TCAYTGATAAGCAGGGCCCAGGG, TTCAYTGATAAGCAGGGCCCAGG	Cohen syndrome, not specified
137852375	NM_000132.3(F8):c.5372T>C (p.Met1791Thr)	TCAYGGTGAGTTAACGGACAGTGG	Hereditary factor VIII deficiency disease
11567847	NM_021961.5(TEAD1):c.1261T>C (p.Tyr?His)	TCATATTYACAGGCTTGAAAGG	
786203989	NM_016069.9(PAM16):c.226A>G (p.Asn76Asp)	CATAGTYCTGCAGAGGAGAGGGG, TCATAGTYCTGCAGAGGAGAGGG	Chondrodysplasia, megarbane-dagher-melki type
587776437	NC_012920.1:m.9478T>C	TCAGAAGYTTTTCTTCGCAGG	Leigh disease
121912474	NM_000424.3(KRT5):c.20T>C (p.Val7Ala)	TCAAGTGYGTCTTCCGGAGCGGG, CAAGTGYGTCTTCCGGAGCGGGG, AAGTGYGTCTTCCGGAGCGGGGG, AGTGYGTCTTCCGGAGCGGGGG	Epidermolysis bullosa simplex, Koebner type
104886461	NM_020533.2(MCOLN1):c.406-2A>G	TACYTGGGAGAGAAGGGGAGG, AGGTACYTGGGAGAGAAGGGG, CAGGTACYTGGGAGAGAAGGG	Ganglioside sialidase deficiency
104894275	NM_000317.2(PTS):c.155A>G (p.Asn52Ser)	TAAYTGTGCCCATGGCCATTGG	6-pyruvoyl-tetrahydropterin synthase deficiency
587777562	NM_015599.2(PGM3):c.737A>G (p.Asn246Ser)	TAAATGAYTGAGTTGCCCTTGG	Immunodeficiency 23
121964906	NM_000027.3(AGA):c.916T>C (p.Cys306Arg)	GTTATAYGTGCCAATGTGACTGG	Aspartylglycosaminuria
28941769	NM_000356.3(TCOF1):c.149A>G (p.Tyr50Cys)	GTGTGTAYAGATGTCCAGAAGGG	Treacher collins syndrome 1
121434464	m.12297T>C	GTCYTAGGCCCAAAATTTGG	Cardiomyopathy, mitochondrial
121908407	NM_054027.4(ANKH):c.143T>C (p.Met48Thr)	GTCGAGAYGCTGGCCAGCTACGG, TCGAGAYGCTGGCCAGCTACGGG	Chondrocalcinosis 2
59151893	NM_000422.2(KRT17):c.275A>G (p.Asn92Ser)	GTCAYTGGTTCTGCATGGTGG, GCGGTCACTGAGGTTCTGCATGG	Pachyonychia congenita type 2
121909499	NM_002427.3(MMP13):c.272T>C (p.Met91Thr)	GTCAYAAAAAGCCAAGATGCGGG, TCAYAAAAAGCCAAGATGCGGG	
61748478	NM_000552.3(VWF):c.2384A>G (p.Tyr795Cys)	GTCAYAGTTCTGGCACGTTTGG	von Willebrand disease type 2N

387906889	NM_006796.2(AFG3L2):c.1847A>G (p.Tyr616Cys)	GTAYAGAGGTATTGTTCTTTGG	Spastic ataxia 5, autosomal recessive
118203907	NM_000130.4(F5):c.5189A>G (p.Tyr1730Cys)	GTAGYAGGCCAAGCCGACAGG	Factor V deficiency
118203945	NM_013319.2(UBIAD1):c.305A>G (p.Asn102Ser)	GTAAGTGYTGACCAAATTACCGG	Schnyder crystalline corneal dystrophy
267607080	NM_005633.3(SOS1):c.1294T>C (p.Trp432Arg)	GGTYGGGAGGGAAAAGACATTGG	Noonan syndrome 4, Rasopathy
137852953	NM_012464.4(TLL1):c.1885A>G (p.Ile629Val)	GGTTAYGGTGCCTTAAGTTGG	Atrial septal defect 6
118203949	NM_013319.2(UBIAD1):c.695A>G (p.Asn232Ser)	GGTGTGTYGGAATGGAGAATGG	Schnyder crystalline corneal dystrophy
137852952	NM_012464.4(TLL1):c.713T>C (p.Val238Ala)	GGGATTGTYGTTCATGAATTGGG	Atrial septal defect 6
41460449	m.3394T>C	GGCYATATACTACAACACTACGCAAAGG	Leber optic atrophy
80357281	NM_007294.3(BRCA1):c.5291T>C (p.Leu1764Pro)	GGGCYAGAAATCTGTGCTATGG, GGCYAGAAATCTGTGCTATGGG	Familial cancer of breast, Breast-ovarian cancer, familial 1
5030764	NM_000174.4(GP9):c.182A>G (p.Asn61Ser)	GGCTGTYGTTGGCCACGAGAAGG	Bernard-Soulier syndrome type C
72556282	NM_000531.5(OTC):c.526T>C (p.Tyr176His)	GGCTGATYACCTCACGCTCCAGG, GATYACCTCACGCTCCAGGTTGG	not provided
121913594	NM_000530.6(MPZ):c.242A>G (p.His81Arg)	GGCATAGYGGAAGATCTATGAGG	Charcot-Marie-Tooth disease type 1B
587777736	NM_017617.3(NOTCH1):c.1285T>C (p.Cys429Arg)	GGCAAGYGCATCAACACGCTGGG, GGCAAGYGCATCAACACGCTGG	Adams-Oliver syndrome 1, Adams-Oliver syndrome 5
63750912	NM_016835.4(MAPT):c.1839T>C (p.Asn613=)	GGATAAYATCAAACACGTCCCCGG, GATAAYATCAAACACGTCCCCGG	Frontotemporal dementia
121918075	NM_000371.3(TTR):c.401A>G (p.Tyr134Cys)	GGAGGYAGGGGCTCAGCAGGGCGG, ATAGGAGYAGGGGCTCAGCAGGG	Amyloidogenic transthyretin amyloidosis
730882063	NM_004523.3(KIF11):c.2547+2T>C	GGAGGYATAACTTTGTAAGTGG	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation
397516156	NM_000257.3(MYH7):c.2546T>C (p.Met849Thr)	GGAGAYGGCCTCCATGAAGGAGG	Primary familial hypertrophic cardiomyopathy, Cardiomyopathy
118204430	NM_000035.3(ALDOB):c.442T>C (p.Trp148Arg)	GGAAGYGGCGTGCTGTGCTGAGG	Hereditary fructosuria
200198778	NM_013382.5(POMT2):c.1997A>G (p.Tyr666Cys)	GGAAGYAGTGGTGGAAAGTAGAGG	Congenital muscular dystrophy, Congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies, type A2, Muscular dystrophy, Congenital muscular dystrophy- dystroglycanopathy with mental retardation, type B2
754896795	NM_004006.2(DMD):c.6982A>T (p.Lys2328Ter)	GCTTTTYTTCAGCTGCCAAGG	Duchenne muscular dystrophy, Becker muscular dystrophy, Dilated cardiomyopathy 3B
148924904	NM_000546.5(TP53):c.488A>G (p.Tyr163Cys)	GCTTGYAGATGGCCATGGCGCGG	Heredity cancer-predisposing syndrome
786204770	NM_016035.4(COQ4):c.155T>C (p.Leu52Ser)	GCTGTYGGCCGCCGGCTCCGCGG	COENZYME Q10 DEFICIENCY, PRIMARY, 7
121909520	NM_001100.3(ACTA1):c.350A>G (p.Asn117Ser)	CGGYTGGCCTGGGATTGAGGG, GCGGYTGGCCTGGGATTGAGGG, CGCGGYTGGCCTGGGATTGAGG	Nemaline myopathy 3
587776879	NM_004656.3(BAP1):c.438-2A>G	GCCYGGGGAAAAACAGAGTCAGG	Tumor predisposition syndrome
727504434	NM_000501.3(ELN):c.890-2A>G	GCCYGAAAACACAGCCCACAGAGG	Supravalvar aortic stenosis
119455953	NM_000391.3(TPP1):c.1093T>C (p.Cys365Arg)	GCCGGGYGTTGGTCTGTCTCTGG	Ceroid lipofuscinosis, neuronal, 2
121964983	NM_000481.3(AMT):c.125A>G (p.His42Arg)	GCCAGGYGGAAGTCATAGACGGG	Non-ketotic hyperglycinemia
121908300	NM_001005741.2(GBA):c.751T>C (p.Tyr251His)	GCCAGAYACTTTGTAAGTAAGGG	Gaucher disease, type 1
786205083	NM_003494.3(DYSF):c.3443-33A>G	GCCAGAGYGAGTGGGAGTGG	Limb-girdle muscular dystrophy, type 2B
121908133	NM_175073.2(APTX):c.602A>G (p.His201Arg)	GCCAAYGGTAACGGGCCCTTGGG, AGCCAAYGGTAACGGGCCCTTGG	Adult onset ataxia with oculomotor apraxia
587777195	NM_005017.3(PCYT1A):c.571T>C (p.Phe191Leu)	GCATGTTGCTCAAACACAGAGG	Spondylometaphyseal dysplasia with cone-rod dystrophy
431905520	NM_014714.3(IFT140):c.4078T>C (p.Cys1360Arg)	CAAGCAGYGTGAGCTGCTCCTGG, GCAGYGTGAGCTGCTCCTGGAGG	Renal dysplasia, retinal pigmentary dystrophy, cerebellar ataxia and skeletal dysplasia
121912889	NM_001844.4(COL2A1):c.4172A>G (p.Tyr1391Cys)	GCAGTGGYAGGTGATGTTCTGGG	Spondyloperipheral dysplasia, Platyspondylitic lethal skeletal dysplasia Torrance type
137854492	NM_001363.4(DKC1):c.1069A>G (p.Thr357Ala)	GCAGGYAGAGATGACCGCTGTGG	Dyskeratosis congenita X-linked
121434362	NM_152783.4(D2HGDH):c.1315A>G (p.Asn439Asp)	GCAGGTYACCATCTCTGGAGGG, TGCAGGTYACCATCTCTGGAGG	D-2-hydroxyglutaric aciduria 1
80338732	NM_002764.3(PRPS1):c.344T>C (p.Met115Thr)	GCAAATAYGCTATCTGTAGCAGG	Charcot-Marie-Tooth disease, X-linked recessive, type 5
387906675	NM_000313.3(PRGS1):c.701A>G (p.Tyr234Cys)	GATTAYATCTGTAGCCTTCGGG, AGATTAYATCTGTAGCCTTCGGG, GAGATTAYATCTGTAGCCTTCGG	Thrombophilia due to protein S deficiency, autosomal recessive

28935478	NM_000061.2(BTK):c.1082A>G (p.Tyr361Cys)	GATGGYAGTTAATGAGCTCAGGG, TGATGGYAGTTAATGAGCTCAGG	
201777056	NM_005050.3(ABCD4):c.956A>G (p.Tyr319Cys)	GATGAGGYAGATGCACACAAAGG	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cblJ TYPE
121918528	NM_000098.2(CPT2):c.359A>G (p.Tyr120Cys)	GATAGGYACATATCAAACCAGGG, AGATAGGYACATATCAAACCAGG	Carnitine palmitoyltransferase II deficiency, infantile
267607014	NM_002942.4(ROBO2):c.2834T>C (p.Ile945Thr)	GAGAYTGGAAATTGGCCGTGG	Vesicoureteral reflux 2
281865192	NM_025114.3(CEP290):c.2991+1655A>G	GATAYTCACAATTACAACACTGGGG, AGATAYTCACAATTACAACACTGGGG, GAGATAYTCACAATTACAACACTGG	Leber congenital amaurosis 10
386833492	NM_000112.3(SLC26A2):c.-26+2T>C	GAGAGGGAGAAGAGGGGAAGCGG	Diastrophic dysplasia
587779773	NM_001101.3(ACTB):c.356T>C (p.Met119Thr)	GAGAAGAGYACCCCAGGTGAGTGG	Baraitser-Winter syndrome 1
121913512	NM_000222.2(KIT):c.1924A>G (p.Lys642Glu)	GAECTTYGAGTTCAGACATGAGGG, GGACTTYGAGTTCAGACATGAGG	
28939072	NM_006329.3(FBLN5):c.506T>C (p.Ile169Thr)	GACAYTGATGAATGTCGCTATGG	Age-related macular degeneration 3
104894248	NM_000525.3(KCNJ11):c.776A>G (p.His259Arg)	GACAYGGTAGATGATCAGCGGGG, TGACAYGGTAGATGATCAGCGGG, ATGACAYGGTAGATGATCAGCGG	Islet cell hyperplasia
387907132	NM_016464.4(TMEM138):c.287A>G (p.His96Arg)	GACAYGAAGGGAGATGCTGAGGG, AGACAYGAAGGGAGATGCTGAGG	Joubert syndrome 16
121918170	NM_000275.2(OCA2):c.1465A>G (p.Asn489Asp)	GACATYTGGAGGGTCCCCGATGG	Tyrosinase-positive oculocutaneous albinism
122467173	NM_014009.3(FOXP3):c.970T>C (p.Phe324Leu)	GACAGAGYTCCTCCACAACATGG	Insulin-dependent diabetes mellitus secretory diarrhea syndrome
137852268	NM_000133.3(F9):c.1328T>C (p.Ile443Thr)	GAAYATATAACCAAGGTATCCGG	Hereditary factor IX deficiency disease
149054177	NM_001999.3(FBN2):c.3740T>C (p.Met1247Thr)	GAATGTAYGATAATGAACGGAGG	not specified, Macular degeneration, early-onset
137854488	NM_212482.1(FN1):c.2918A>G (p.Tyr973Cys)	GAAGTAAYAGGTGACCCCCAGGGG	Glomerulopathy with fibronectin deposits 2
786204027	NM_005957.4(MTHFR):c.1530+2T>C	GAAGGYGTGGTAGGGAGGCACGG, AAGGYGTGGTAGGGAGGCACGGG, AGGYGTGGTAGGGAGGCACGGG	Homocystinemia due to MTHFR deficiency
104894223	NM_012193.3(FZD4):c.766A>G (p.Ile256Val)	GAAATAYGATGGGGCGCTCAGGG, AGAAATAYGATGGGGCGCTCAGG	Retinopathy of prematurity
137854474	NM_000138.4(FBN1):c.3793T>C (p.Cys1265Arg)	CTTGYGTTATGATGGATTATGG	Marfan syndrome
587784418	NM_006306.3(SMC1A):c.3254A>G (p.Tyr1085Cys)	CTTAYAGATCTCATCAATGTTGG	Congenital muscular hypertrophy-cerebral syndrome
81002805	NM_000059.3(BRCA2):c.316+2T>C	CTTAGGYAAGTAATGCAATATGG	Familial cancer of breast, Breast-ovarian cancer, familial 2, Hereditary cancer-predisposing syndrome
121909653	NM_182925.4(FLT4):c.3104A>G (p.His1035Arg)	CTGYGGATGCACTGGGGTGCAGG, TCTGYGGATGCACTGGGGTGCAGG	
786205107	NM_031226.2(CYP19A1):c.743+2T>C	CTGTGYAAGTAATACAACATTGG	Aromatase deficiency
587777037	NM_001283009.1(RTEL1):c.3730T>C (p.Cys1244Arg)	CTGTGTGYGCCAGGGCTGTGGGG	Dyskeratosis congenita, autosomal recessive, 5
794728380	NM_000238.3(KCNH2):c.1945+6T>C	CTGTGAGYGTGCCAGGGGGCGGG, TGAGYGTGCCAGGGGGCGGG	Cardiac arrhythmia
267607987	NM_000251.2(MSH2):c.2005+2T>C	CTGGYAAAAAACCTGTTTGG, TGGYAAAAAACCTGTTTGG	Hereditary Nonpolyposis Colorectal Neoplasms
397509397	NM_006876.2(B4GAT1):c.1168A>G (p.Asn390Asp)	TGATYTTCAGCCTCTTTGGGG, CTGATYTTCAGCCTCTTTGGGG, GCTGATYTTCAGCCTCTTTGGGG	Congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies, type A13
121918381	NM_000040.1(APOC3):c.280A>G (p.Thr94Ala)	CTGAAGYTGGTCTGACCTCAGGG, GCTGAAGYTGGTCTGACCTCAGGG	
104894919	NM_001015877.1(PHF6):c.769A>G (p.Arg257Gly)	CTCYTGATGTTGTTGAGCTGG	Borjeson-Forssman-Lehmann syndrome
267606869	NM_005144.4(HR):c.-218A>G	CTCYAGGGCCGCAGGTTGGAGGG, GCTCYAGGGCCGCAGGTTGGAGGG, GGCCTCYAGGGCCGCAGGTTGG	Marie Unna hereditary hypotrichosis 1
139732572	NM_000146.3(FTL):c.1A>G (p.Met1Val)	CTCAYGGTGGTTGGCAAGAAGG	L-ferritin deficiency
397515418	NM_018486.2(HDAC8):c.1001A>G (p.His334Arg)	CTCAYGATCTGGGATCTCAGAGG	Cornelia de Lange syndrome 5
372395294	NM_198056.2(SCN5A):c.1247A>G (p.Tyr416Cys)	CTCAYAGGCCATTGCGACCACGG	not provided
104895304	NM_000431.3(MVK):c.803T>C (p.Ile268Thr)	CTCAAYAGATGCCATCTCCCTGG	Hyperimmunoglobulin D with periodic fever, Mevalonic aciduria
587777188	NM_001165899.1(PDE4D):c.1850T>C (p.Ile617Thr)	CTATAYTGTTCATCCCCCTCTGG, ACTATAYTGTTCATCCCCCTCTGG	Acrodysostosis 2, with or without hormone resistance
398123026	NM_003867.3(FGF17):c.560A>G (p.Asn187Ser)	CGTGGYTGGGAAGGGCAGCTGG	Hypogonadotropic hypogonadism 20 with or without anosmia

121964924	NM_001385.2(DPYS):c.1078T>C (p.Trp360Arg)	CGTAATAYGGGAAAAGGCCTGG, AATAYGGGAAAAGGCCTGGTGG, ATAYGGGAAAAGGCCTGGTGGG	Dihydropyrimidinase deficiency
587777301	NM_199189.2(MATR3):c.1864A>G (p.Thr622Ala)	CGGYTGAACTCTCAGTCTTCTGG	Myopathy, distal, 2
200238879	NM_000527.4(LDLR):c.694+2T>C	ACTGCGGYATGGCGGGGCCAGGG, CTGCGGYATGGCGGGGCCAGGG, CGGYATGGCGGGGCCAGGGTGG	Familial hypercholesterolemia
142951029	NM_145046.4(CALR3):c.245A>G (p.Lys82Arg)	CGGTYTGAAGCGTGCAGAGATGG	Arrhythmogenic right ventricular cardiomyopathy, Familial hypertrophic cardiomyopathy 19, Hypertrophic cardiomyopathy
786200953	NM_006785.3(MALT1):c.1019-2A>G	CGCYTTGAAAAAAAAGAAAGGG, TCGCTTGAAAAAAAAGAAAGG	Combined immunodeficiency
120074192	NM_000218.2(KCNQ1):c.418A>G (p.Ser140Gly)	CGCYGAAGATGAGGCAGACCAAGG	Atrial fibrillation, familial, 3, Atrial fibrillation
267606887	NM_005957.4(MTHFR):c.971A>G (p.Asn324Ser)	CGCGGYTGAGGGTAGAAAGTGG	Homocystinuria due to MTHFR deficiency
118192117	NM_000540.2(RYR1):c.1205T>C (p.Met402Thr)	CGCACGATCCACAGCACCAATGG	Congenital myopathy with fiber type disproportion, Central core disease
199473625	NM_198056.2(SCN5A):c.4978A>G (p.Ile1660Val)	CGAYGTTGAAGAGGGCAGGCAGG, AGCCCAGYGTGAAGAGGGCAGG	Brugada syndrome
794726865	NM_000921.4(PDE3A):c.1333A>G (p.Thr445Ala)	CGAGGGYGGTGGTGGTCCAAGTGG	Brachydactyly with hypertension
606231254	NM_005740.2(DNAL4):c.153+2T>C	CGAGGYATTGCCAGCAGTGCAGG	Mirror movements 3
786204826	NM_004771.3(MMP20):c.611A>G (p.His204Arg)	CGAAAYGTGTATCTCCTCCCAAGG	Amelogenesis imperfecta, hypomaturation type, IIA2
796053139	NM_021007.2(SCN2A):c.4308+2T>C	CGAAATGYAAGTCTAGTTAGAGG, GAAATGYAAGTCTAGTTAGAGGG	not provided
137854494	NM_005502.3(ABCA1):c.4429T>C (p.Cys1477Arg)	CCTGTGYGTCCCCCAGGGGCAGGG, CTGTGYGTCCCCCAGGGGCAGGG, TGTGYGTCCCCCAGGGGCAGGGGG, GTGYGTCCCCCAGGGGCAGGGGG	Tangier disease
786205144	NM_001103.3(ACTN2):c.683T>C (p.Met228Thr)	CCTAAAYGTTGGATGCTGAAGG	Dilated cardiomyopathy 1AA
199919568	NM_007254.3(PNKP):c.1029+2T>C	CCGGYGAGGCCCTGGGGCGGGGG, TCCGGYGAGGCCCTGGGGCGGGGG, ATCCGGYGAGGCCCTGGGGCGGGG, GATCCGGYGAGGCCCTGGGGCGGG	not provided
28939079	NM_018965.3(TREM2):c.401A>G (p.Asp134Gly)	TGAYCAGGGGGTCTATGGGAGG, CGGTGAYCCAGGGGGTCTATGGG, CCGGTGTGAYCCAGGGGGTCTATGG	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy
193302855	NM_032520.4(GNPTG):c.610-2A>G	CCCYGAAGGTGGAGGATGCAGGG, GCCCYGAAGGTGGAGGATGCAGGG	Mucolipidosis III Gamma
111033708	NM_000155.3(GALT):c.499T>C (p.Trp167Arg)	CCCTYGGGTGCAGGTTGTGAGG	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
28933378	NM_000174.4(GP9):c.70T>C (p.Cys24Arg)	CCCAYGTAACCTGCCGCCCTGG	Bernard Soulier syndrome, Bernard-Soulier syndrome type C
364897	NM_000157.3(GBA):c.680A>G (p.Asn227Ser)	CCAYTGGTCTTGAGCCAAGTGGG, TCCAYTGGTCTTGAGCCAAGTGG	Gaucher disease, Subacute neuronopathic Gaucher disease, Gaucher disease, type 1
796052551	NM_000833.4(GRIN2A):c.2449A>G (p.Met817Val)	CCAYTGTCAATGCCAGCTGG	not provided
63751006	NM_002087.3(GRN):c.2T>C (p.Met1Thr)	CCAYGTGGACCCTGGTGAGCTGG	Frontotemporal dementia, ubiquitin-positive
786203997	NM_001031.4(RPS28):c.1A>G (p.Met1Val)	TGTCCAYGATGGCGGGCGGGCGG, CCAYGATGGCGGGCGGGCGGGCGG	Diamond-Blackfan anemia with microtia and cleft palate
121908595	NM_002755.3(MAP2K1):c.389A>G (p.Tyr130Cys)	CCAYAGAACCCCACGATGTACGG	Cardiofaciocutaneous syndrome 3, Rasopathy
398122910	NM_000431.3(MVK):c.1039+2T>C	CCAGGYATCCCGGGGGTAGGTGG, CAGGYATCCCGGGGGTAGGTGG	Porokeratosis, disseminated superficial actinic 1
119474039	NM_020365.4(EIF2B3):c.1037T>C (p.Ile346Thr)	CCAGAYTGTCAACACCTGG	Leukoencephalopathy with vanishing white matter
587777866	NM_000076.2(CDKN1C):c.*5+2T>C	CCAAGYGAGTACAGCCACCTGG, CAAGYGAGTACAGCCACCTGGG, AAGYGAGTACAGCGCACCTGGGG	Beckwith-Wiedemann syndrome
121918530	NM_005587.2(MEF2A):c.788A>G (p.Asn263Ser)	AGAYTACCAACCTGGTGGAGG, CCAAGAYTACCAACCTGGTGG	
483352818	NM_000211.4(ITGB2):c.1877+2T>C	CATGYGAGTGCAGGGGGAGCAGG	Leukocyte adhesion deficiency type 1
460184	NM_000186.3(CFH):c.3590T>C (p.Val1197Ala)	CAGYTAATTGTGTAAACGG	Atypical hemolytic-uremic syndrome 1
121908423	NM_004795.3(KL):c.578A>G (p.His193Arg)	CAGYGGTACAGGGTGACCACGGG, CCAGYGGTACAGGGTGACCACGG	
281860300	NM_005247.2(FGF3):c.146A>G (p.Tyr49Cys)	CAGYAGAGCTTGCAGGGCCGGGG, GCAGYAGAGCTTGCAGGGCCGGGG, CGCAGYAGAGCTTGCAGGGCCGGGG	Deafness with labyrinthine aplasia microtia and microdontia (LAMM)
28935488	NM_000169.2(GLA):c.806T>C (p.Val269Ala)	CAGTTAGYGATTGGCAACTTGG	Fabry disease

587776514	NM_173560.3(RFX6):c.380+2T>C	CAGTGGYGAGACTCGCCCGCAGG, AGTGGYGAGACTCGCCCGCAGGG	Mitchell-Riley syndrome
104894117	NM_178138.4(LHX3):c.332A>G (p.Tyr111Cys)	CAGGTGGYACACGAAGTCCTGGG	Pituitary hormone deficiency, combined 3
34878913	NM_000184.2(HBG2):c.125T>C (p.Phe42Ser)	CAGAGGYCTTTGACAGCTTG	Cyanosis, transient neonatal
120074124	NM_000543.4(SMPD1):c.911T>C (p.Leu304Pro)	AGCACYTGTGAGGAAGTCCCTGG, GCACYTGTGAGGAAGTCCCTGGG, CACYTGTGAGGAAGTCCCTGGGG	Sphingomyelin/cholesterol lipidosis, Niemann-Pick disease, type A, Niemann-Pick disease, type B
281860272	NM_005211.3(CSF1R):c.2320-2A>G	CACYGAGGGAAAGCAGTCAGGG, GCACYGAGGGAAAGCAGTCAGGG	Heredity diffuse leukoencephalopathy with spheroids
128624216	NM_000033.3(ABCD1):c.443A>G (p.Asn148Ser)	CACTGTYGACGAAGGTAGCAGGG, GCACTGTYGACGAAGGTAGCAGGG	Adrenoleukodystrophy
398124257	NM_012463.3(ATP6V0A2):c.825+2T>C	CACTGYGAGTAAGCTGAAAGTGG	Cutis laxa with osteodystrophy
267606679	NM_004183.3(BEST1):c.704T>C (p.Val235Ala)	CACTGGYGTATACACAGGTGAGG	Vitreoretinochoroidopathy dominant
397514518	NM_000344.3(SMN1):c.388T>C (p.Tyr130His)	CACTGGAYATGGAATAGAGAGG	Kugelberg-Welander disease
143946794	NM_001946.3(DUSP6):c.566A>G (p.Asn189Ser)	CACTAYGGGTCTCGGTCAAGG	Hypogonadotropic hypogonadism 19 with or without anosmia
397516076	NM_000256.3(MYBPC3):c.821+2T>C	GCACGYGAGTGGCCATCCTCAGG, CACGYGAGTGGCCATCCTCAGGG	Familial hypertrophic cardiomyopathy 4, not specified
149977726	NM_001257988.1(TYMP):c.665A>G (p.Lys222Arg)	CACGAGTYTCTTACTGAGAATGG, GAGTYTCTTACTGAGAATGGAGG	
121917770	NM_003361.3(UMOD):c.383A>G (p.Asn128Ser)	CACAYTGACACATGTGGCCAGGG, CCACAYTGACACATGTGGCCAGGG	Familial juvenile gout
121909008	NM_000492.3(CFTR):c.2738A>G (p.Tyr913Cys)	CACATAAYACGAACCTGGTGCTGG	Cystic fibrosis
137852819	NM_003688.3(CASK):c.2740T>C (p.Trp914Arg)	CACAGYGGGTCCCTGTCCCTGG, ACAGYGGGTCCCTGTCCCTGGG	FG syndrome 4
74315320	NM_024009.2(GJB3):c.421A>G (p.Ile141Val)	CAAYGATGAGCTTGAAGATGAGG	Deafness, autosomal recessive
80356747	NM_001701.3(BAAT):c.967A>G (p.Ile323Val)	CAAYGAAGAGGAATTGCCCTGG	Atypical hemolytic-uremic syndrome 1
180177324	NM_012203.1(GRHPR):c.934A>G (p.Asn312Asp)	CAAGTYGTTAGCTGCCAACAGG	Primary hyperoxaluria, type II
281860274	NM_005211.3(CSF1R):c.2381T>C (p.Ile794Thr)	CAAGAYTGGGACTTCGGGCTGG	Heredity diffuse leukoencephalopathy with spheroids
398122908	NM_005334.2(HCF1C):c.-970T>C	CAAGAYGGCGGCTCCAGGGAGG	Mental retardation 3, X-linked
548076633	NM_002693.2(POLG):c.3470A>G (p.Asn1157Ser)	CAAGAGGYTGGTGATCTGCAAGG	not provided
120074146	NM_000019.3(ACAT1):c.935T>C (p.Ile312Thr)	CAAGAAYAGTAGGTAAGGCCAGG	Deficiency of acetyl-CoA acetyltransferase
397514489	NM_005340.6(HINT1):c.250T>C (p.Cys84Arg)	CAAGAAAYGTGCTGCTGATCTGG, AAGAAAYGTGCTGCTGATCTGGG	Gamstorp-Wohlfart syndrome
587783539	NM_178151.2(DCX):c.2T>C (p.Met1Thr)	CAAAATAYGGAACCTTGATTTGG	Heterotopia
104894765	NM_005448.2(BMP15):c.704A>G (p.Tyr235Cys)	ATTGAAAYAGAGTAACAAGAAGG	Ovarian dysgenesis 2
137852429	NM_000132.3(F8):c.1892A>G (p.Asn631Ser)	ATGYTGGAGGCTTGGAACTCTGG	Heredity factor VIII deficiency disease
72558441	NM_000531.5(OTC):c.779T>C (p.Leu260Ser)	ATGTATYAATTACAGACACTTGG	not provided
398123765	NM_003494.3(DYSF):c.1284+2T>C	ATGGYAAGGAGCAAGGGAGCAGG	Limb-girdle muscular dystrophy, type 2B
387906924	NM_020191.2(MRPS22):c.644T>C (p.Leu215Pro)	ATCYTAGGGTAAGGTGACTTAGG	Combined oxidative phosphorylation deficiency 5
397518039	NM_206933.2(USH2A):c.8559-2A>G	ATCYAAAGCAAAGACAAGCAGG	Retinitis pigmentosa, Usher syndrome, type 2A
5742905	NM_000071.2(CBS):c.833T>C (p.Ile278Thr)	ATCAYTGGGGTGGATCCCGAAGGG, TCAYTGGGGTGGATCCCGAAGGG	Homocystinuria due to CBS deficiency, Homocystinuria, pyridoxine-responsive
397507473	NM_004333.4(BRAF):c.1403T>C (p.Phe468Ser)	ATCATYGGAACAGCTCTACAAGG, TCATYGGAACAGCTCTACAAGGG	Cardiofaciocutaneous syndrome, Rasopathy
786204056	NM_000264.3(PTCH1):c.3168+2T>C	ATCATTGAGGTGTATTATAAGGG, CATTGAGGTGTATTATAAGGGG	Gorlin syndrome
72558484	NM_000531.5(OTC):c.1005+2T>C	ATCATGGYAAGCAAGAAACAAGG	not provided
199473074	NM_000335.4(SCN5A):c.688A>G (p.Ile230Val)	ATAYAGTTTCAGGGCCGGAGGG, CTGATAYAGTTTCAGGGCCGG	Brugada syndrome
111033273	NM_206933.2(USH2A):c.1606T>C (p.Cys536Arg)	ATATAGAYGCCTCTGCTCCCAGG	Usher syndrome, type 2A
72556290	NM_000531.5(OTC):c.542A>G (p.Glu181Gly)	ATAGTGYCCTAAAAGGCACGGG	not provided
121918711	NM_004612.3(TGFBR1):c.1199A>G (p.Asp400Gly)	ATAGATGYCAGCACGTTGAGG	Loeys-Dietz syndrome 1
104886288	NM_000495.4(COL4A5):c.4699T>C (p.Cys1567Arg)	AGTAYGTGAAGCTCCAGCTGTGG	Alport syndrome, X-linked recessive

144637717	NM_016725.2(FOLR1):c.493+2T>C	CTTCAGGGYAGGGCTGGGGTGGG, AGGYAGGGCTGGGTGGGCAGG	not provided
72558492	NM_000531.5(OTC):c.1034A>G (p.Tyr345Cys)	AGGTGAGYAATCTGTCAGCAGGG	not provided
62638745	NM_000121.3(EPOR):c.1460A>G (p.Asn487Ser)	AGGGYTGGAGTAGGGGCCATCGG	Acute myeloid leukemia, M6 type, Familial erythrocytosis, 1
387907021	NM_031427.3(DNAL1):c.449A>G (p.Asn150Ser)	AGGGAYTGCTACAAACACCAGG	Kartagener syndrome, Ciliary dyskinesia, primary, 16
397514488	NM_001161581.1(POC1A):c.398T>C (p.Leu133Pro)	AGCYGTGGACAAGAGCAGCCGG	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis
154774633	NM_017882.2(CLN6):c.200T>C (p.Leu67Pro)	AGCYGGTATTCCCCTCTCGAGTGG	Adult neuronal ceroid lipofuscinosis
111033700	NM_000155.3(GALT):c.482T>C (p.Leu161Pro)	AGCYGGGTGCCAGTACCCCTGG	Deficiency of UDPglucose-hexose-1-phosphate uridyltransferase
128621198	NM_000061.2(BTK):c.1223T>C (p.Leu408Pro)	GAGCYGGGACTGGACAATTGGG, AGCYGGGACTGGACAATTGGG	X-linked agammaglobulinemia
137852611	NM_000211.4(ITGB2):c.446T>C (p.Leu149Pro)	AGCYAGGTGGCGACTGCTCCGG	Leukocyte adhesion deficiency
121908838	NM_003722.4(TP63):c.697A>G (p.Lys233Glu)	AGCTTYTTGTAGACAGGCATGG	Split-hand/foot malformation 4
397515869	NM_000169.2(GLA):c.1153A>G (p.Thr385Ala)	AGCTGTGYGATGAAGCAGGCAGG	not specified
118204064	NM_000237.2(LPL):c.548A>G (p.Asp183Gly)	GCTGGAYCGAGGCCCTAAAAGGG, AGCTGGAYCGAGGCCCTAAAAGGG	Hyperlipoproteinemia, type I
128620186	NM_000061.2(BTK):c.2T>C (p.Met1Thr)	AGCTAYGGCCGCAGTGATTCTGG	X-linked agammaglobulinemia
786204132	NM_014946.3(SPAST):c.1165A>G (p.Thr389Ala)	ATTGYCTTCCCATTCCCAGGTGG, AGCATTYGCTTCCCATTCCCAGG	Spastic paraparesis 4, autosomal dominant
199473661	NM_000218.2(KCNQ1):c.550T>C (p.Tyr184His)	CAGCAAGBACGTGGGCCCTCTGGG, AGCAAGBACGTGGGCCCTCTGGG, GCAAGBACGTGGGCCCTCTGGGG	Congenital long QT syndrome, Cardiac arrhythmia
387907129	NM_024599.5(RHBD2):c.557T>C (p.Ile186Thr)	AGAYTGTGGATCCGCTGGCCCGG	Howell-Evans syndrome
387906702	NM_006306.3(SMC1A):c.2351T>C (p.Ile784Thr)	AGAYTGGTGTGCGAACATCCGG	Congenital muscular hypertrophy-cerebral syndrome
193929348	NM_000525.3(KCNJ11):c.544A>G (p.Ile182Val)	AGAYGAGGGTCTAGCCCTCGGG	Permanent neonatal diabetes mellitus
121908934	NM_004086.2(COCH):c.1535T>C (p.Met512Thr)	AGATAYGGCTCTAACCGAAGG	Deafness, autosomal dominant 9
397514377	NM_000060.3(BTD):c.641A>G (p.Asn214Ser)	AGAGGYTGTGTTACGGTAGCGG	Biotinidase deficiency
72552295	NM_000531.5(OTC):c.2T>C (p.Met1Thr)	AGAAGAYGCTGTTAACATCGAGG	not provided
201893545	NM_016247.3(IMPG2):c.370T>C (p.Phe124Leu)	ACTYTTGGATCGACTTCCCTGG	Macular dystrophy, vitelliform, 5
121434469	m.4290T>C	ACTYTGATAGAGTAAATAATAGG	
121918733	NM_006920.4(SCN1A):c.269T>C (p.Phe90Ser)	ACTTYTATAGTATTGAATAAAGG, CTTYTATAGTATTGAATAAAGG	Severe myoclonic epilepsy in infancy
121434471	m.4291T>C	ACTTYGATAGAGTAAATAATAGG	Hypertension, hypercholesterolemia, and hypomagnesemia, mitochondrial
606231289	NM_001302946.1(TRNT1):c.497T>C (p.Leu166Ser)	ACTTYATTTGACTACTTAATGG	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay
63750067	NM_000517.4(HBA2):c.*92A>G	CTTYATTCAAAGACCAGGAAGGG, ACTTYATTCAAAGACCAGGAAGG	Hemoglobin H disease, nondeletional
121918734	NM_006920.4(SCN1A):c.272T>C (p.Ile91Thr)	ACTTTTAYAGTATTGAATAAAGG, CTTTTAYAGTATTGAATAAAGG	Severe myoclonic epilepsy in infancy
137854557	NM_000267.3(NF1):c.1466A>G (p.Tyr489Cys)	ACTTAYAGCTTCTGTCTCCAGG	Neurofibromatosis, type 1
397514626	NM_018344.5(SLC29A3):c.607T>C (p.Ser203Pro)	ACTGATAYCAGGTGAGAGCCAGG, CTGATAYCAGGTGAGAGCCAGGG	Histiocytosis-lymphadenopathy plus syndrome
118204440	NM_000512.4(GALNS):c.1460A>G (p.Asn487Ser)	ACGYTGAGCTGGGCCCTGCAGGG, CACGYTGAGCTGGGCCCTGCAGGG	Mucopolysaccharidosis, MPS-IV-A
587776843	NG_012088.1:g.2209A>G	ACCYTATGATCCGCCGCCCTTGG	
137853033	NM_001080463.1(DYNC2H1):c.4610A>G (p.Gln1537Arg)	ACCYGTGAAGGGAAACAGAGATGG	Short-rib thoracic dysplasia 3 with or without polydactyly
28933698	NM_000435.2(NOTCH3):c.1363T>C (p.Cys455Arg)	TTCACCGTATCTGTATGGCAGG, ACCGTGTATCTGTATGGCAGG	Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy
587776766	NM_000463.2(UGT1A1):c.1085-2A>G	ACCYGAGATGCAAATAGGGAGG, GTGACCGAGATGCAAATAGGGAGG	Crigler Najjar syndrome, type 1
587781628	NM_001128425.1(MUTYH):c.1187-2A>G	ACCYGAGAGGGAGGGCAGCCAGG	Hereditary cancer-predisposing syndrome, Carcinoma of colon
61755817	NM_000322.4(PRPH2):c.736T>C (p.Trp246Arg)	ACCTGYGGGTGCGTGGCTGCAGGG, CCTGYGGGTGCGTGGCTGCAGGG	Retinitis pigmentosa
121909184	NM_001089.2(ABC3A):c.1702A>G (p.Asn568Asp)	ACCGTYGTGGCCCAGCAGGACGG	Surfactant metabolism dysfunction, pulmonary, 3

121434466	m.4269A>G	ACAYATTTCTTAGGTTGAGGGG, GACAYATTCTTAGGTTGAGGG, AGACAYATTCTTAGGTTGAGG	
794726768	NM_001165963.1(SCN1A):c.1048A>G (p.Met350Val)	ACAYATATCCCTCTGGACATTGG	Severe myoclonic epilepsy in infancy
28934876	NM_001382.3(DPAGT1):c.509A>G (p.Tyr170Cys)	ACAYAGTACAGGATTCTGCAGGG, GACAYAGTACAGGATTCTGCAGGG	Congenital disorder of glycosylation type 1J
104894749	NM_000054.4(AVPR2):c.614A>G (p.Tyr205Cys)	ACAYAGGTGCGACGGCCCCAGGG, GACAYAGGTGCGACGGCCCCAGG	Nephrogenic diabetes insipidus, Nephrogenic diabetes insipidus, X-linked
128621205	NM_000061.2(BTK):c.1741T>C (p.Trp581Arg)	ACATTYGGGCTTTGGTAAGTGG	X-linked agammaglobulinemia
28940892	NM_000529.2(MC2R):c.761A>G (p.Tyr254Cys)	ACATGYACCAGGCCAGTAGGGG, GACATGYAGCAGGCCAGTAGGG, AGACATGYAGCAGGCCAGTAGG	ACTH resistance
794726844	NM_001165963.1(SCN1A):c.1046A>G (p.Tyr349Cys)	ACATAYATCCCTCTGGACATTGG	Severe myoclonic epilepsy in infancy
587783083	NM_003159.2(CDKL5):c.449A>G (p.Lys150Arg)	ACAGTYTTAGGACATCATTGTGG	not provided
397514651	NM_000108.4(DLD):c.140T>C (p.Ile47Thr)	ACAGTTAYAGGTTCTGGTCTGG, GTTAYAGGTTCTGGTCTGGAGG	Maple syrup urine disease, type 3
794727060	NM_001848.2(COL6A1):c.957+2T>C	ACAAGGYGAGCGTGGCTGCTGG, CAAGGYGAGCGTGGCTGCTGG	Ullrich congenital muscular dystrophy, Bethlem myopathy
72554346	NM_000531.5(OTC):c.284T>C (p.Leu95Ser)	ACAAGATYGTCTACAGAAACAGG	not provided
483353031	NM_002136.2(HNRNPA1):c.841T>C (p.Phe281Leu)	AATYTTGGAGGCAGAACAGCTGG	Chronic progressive multiple sclerosis
104894271	NM_000315.2(PTH):c.52T>C (p.Cys18Arg)	AATTYGTTTCTTACAAAATCGG	Hypoparathyroidism familial isolated
267608260	NM_015599.2(PGM3):c.248T>C (p.Leu83Ser)	AATGTYGGCACCATCCTGGGAGG	Immunodeficiency 23
267606900	NM_018109.3(MTPAP):c.1432A>G (p.Asn478Asp)	AATGGATYCTGAATGTACAGAGG	Ataxia, spastic, 4, autosomal recessive
796053169	NM_021007.2(SCN2A):c.387-2A>G	AATAAAAGYAGAAATATCGTCAAGG	not provided
104894937	NM_000116.4(TAZ):c.352T>C (p.Cys118Arg)	AAGYGTGTGCCCTGTGCCAGG	3-Methylglutaconic aciduria type 2
104893911	NM_001018077.1(NR3C1):c.1712T>C (p.Val571Ala)	AAGYGATTGCAGCAGTGAAATGG	Pseudohermaphroditism, female, with hypokalemia, due to glucocorticoid resistance
397514472	NM_004813.2(PEX16):c.992A>G (p.Tyr331Cys)	AAGYAGATTTCTGCCAGGTGGG, GAAGYAGATTTCTGCCAGGTGG, GTAGAAGYAGATTTCTGCCAGG	Peroxisome biogenesis disorder 8B
121918407	NM_001083112.2(GPD2):c.1904T>C (p.Phe635Ser)	AAGTYTGATGCAGACAGAAAGG	Diabetes mellitus type 2
63751110	NM_000251.2(MSH2):c.595T>C (p.Cys199Arg)	AAGGAAYGTGTTTACCCGGAGG	Hereditary Nonpolyposis Colorectal Neoplasms
119450945	NM_000026.2(ADSL):c.674T>C (p.Met225Thr)	AAGAYGGTACAGAAAAGGCAGG	Adenylosuccinate lyase deficiency
113993988	NM_002863.4(PYGL):c.2461T>C (p.Tyr821His)	AAGAAAYATGCCCAAAACATCTGG	Glycogen storage disease, type VI
119485091	NM_022041.3(GAN):c.1268T>C (p.Ile423Thr)	AAGAAAAYCTACGCCATGGGTGGAGG	Giant axonal neuropathy
137852419	NM_000132.3(F8):c.1660A>G (p.Ser554Gly)	AACYAGAGTAATAGCGGGTCAGG	Hereditary factor VIII deficiency disease
121964967	NM_000071.2(CBS):c.1150A>G (p.Lys384Glu)	AACTYGGCCTGCAGGGATGGGG, GAACTYGGCCTGCAGGGATGGGG, GGAACACTYGGCCTGCAGGGATGGGG, AGGAACACTYGGCCTGCAGGGATGG	Homocystinuria, pyridoxine-responsive
137852376	NM_000132.3(F8):c.1754T>C (p.Ile585Thr)	AACAGAYAATGTCAGACAAGAGG	Hereditary factor VIII deficiency disease
121917930	NM_006920.4(SCN1A):c.3577T>C (p.Trp1193Arg)	AACAAYGGTGGAACCTGAGAAGG	Generalized epilepsy with febrile seizures plus, type 1, Generalized epilepsy with febrile seizures plus, type 2
28939717	NM_003907.2(EIF2B5):c.271A>G (p.Thr91Ala)	AAATGYTCCTGTACACCTGTGG	Leukoencephalopathy with vanishing white matter
80357276	NM_007294.3(BRCA1):c.122A>G (p.His41Arg)	AAATATGYGGTCACACTTGAGG	Familial cancer of breast, Breast-ovarian cancer, familial 1
397515897	NM_000256.3(MYBPC3):c.1351+2T>C	AAAGGYGGCCTGGGACCTGAGG	Familial hypertrophic cardiomyopathy 4, Cardiomyopathy
397514491	NM_005340.6(HINT1):c.152A>G (p.His51Arg)	AAAAYGTGTTGGTGCTTGAGGGG, GAAAAYGTGTTGGTGCTTGAGGG, AGAAAAYGTGTTGGTGCTTGAGG	Gamstorp-Wohlfart syndrome
387907164	NM_020894.2(UVSSA):c.94T>C (p.Cys32Arg)	AAATTYGCAAGTATGTCTTAGG, AAATTYGCAAGTATGTCTTAGG	UV-sensitive syndrome 3
118161496	NM_025152.2(NUBPL):c.815-27T>C	TGGTCYAATGGATGTCTGCTGG, GGTCYAATGGATGTCTGCTGG	Mitochondrial complex I deficiency
764313717	NM_005609.2(PYGM):c.425_528del	TGGCTGYCAGGGACCCAGCAAGGAGG	
28934568	NM_003242.5(TGFBR2):c.923T>C (p.Leu308Pro)	AGTTCCYGACGGCTGAGGAGCGG	Loeys-Dietz syndrome 2

121913461	NM_007313.2(ABL1):c.814T>C (p.Tyr272His)	CCAGYACGGGGAGGTGTACGAGG, CAGYACGGGGAGGTGTACGAGG	
377750405	NM_173551.4(ANKS6):c.1322A>G (p.Gln441Arg)	AGGGCYGTCGGACCTTCGAGTGG, GGGCYGTCGGACCTTCGAGTGGGG	Nephronophthisis 16
57639980	NM_001927.3(DES):c.1034T>C (p.Leu345Pro)	ATTCYYGATGAGGCAGATGCCG, TTCCCYGATGAGGCAGATGCCGG	Myofibrillar myopathy 1
147391618	NM_020320.3(RARS2):c.35A>G (p.Gln12Arg)	ATACCYGGCAAGCAATAGCGCG	Pontocerebellar hypoplasia type 6
182650126	NM_002977.3(SCN9A):c.2215A>G (p.Ile739Val)	GTAAYTGCAAGATCTACAAAAGG	Small fiber neuropathy
80358278	NM_004700.3(KCNQ4):c.842T>C (p.Leu281Ser)	ACATYGACAACCATCGGCTATGG	DFNA 2 Nonsyndromic Hearing Loss
786204012	NM_005957.4(MTHFR):c.388T>C (p.Cys130Arg)	GACCYGCTGCCGTACGCCCTGG	Homocysteinemia due to MTHFR deficiency
786204037	NM_005957.4(MTHFR):c.1883T>C (p.Leu628Pro)	TCCCACYGGACAACACTGCCTCTGG	Homocysteinemia due to MTHFR deficiency
202147607	NM_000140.3(FECH):c.1137+3A>G	GTAGAYACCTTAGAGAACAAATGG	Erythropoietic protoporphyrina
122456136	NM_005183.3(CACNA1F):c.2267T>C (p.Ile756Thr)	TGCCAYTGCTGTGGACAACCTGG	
786204851	NM_007374.2(SIX6):c.110T>C (p.Leu37Pro)	GTCGYGCGCGTGGCCCCCTGCCG	Cataract, microphthalmia and nystagmus
794728167	NM_000138.4(FBN1):c.1468+2T>C	ATTGGYACGTGATCCATCCTAGG	Thoracic aortic aneurysms and aortic dissections
121964909	NM_000027.3(AGA):c.214T>C (p.Ser72Pro)	GACGGCYCTGTAGGCTTGGAGG	Aspartylglycosaminuria
121964978	NM_000170.2(GLDC):c.2T>C (p.Met1Thr)	CGGCCAYGCAGTCTGTGCCAGG, GGCCAYGCAGTCCTGTGCCAGGG	Non-ketotic hyperglycinemia
121965008	NM_000398.6(CYB5R3):c.446T>C (p.Leu149Pro)	CTGCGGTCTACCAGGGCAAAGG	METHEMOGLOBINEMIA, TYPE I
121965064	NM_000128.3(F11):c.901T>C (p.Phe301Leu)	TGATYTCTTGGGAGAAGAACTGG	Hereditary factor XI deficiency disease
45517398	NM_000548.3(TSC2):c.5150T>C (p.Leu1717Pro)	GCCCYGCACGCAAATGTGAGTGG, CCCYGCACGCAAATGTGAGTGGG	Tuberous sclerosis syndrome
786205857	NM_015662.2(IFT172):c.770T>C (p.Leu257Pro)	TTGTGTYAGGAAGTTATGACAGG	RETINITIS PIGMENTOSA 71
786205904	NM_001135669.1(XPR1):c.653T>C (p.Leu218Ser)	GCGTTYACGTGTCCCCCCTTTGG, CGTTYACGTGTCCCCCCTTTGGG	BASAL GANGLIA CALCIFICATION, IDIOPATHIC, 6
104893704	NM_000388.3(CASR):c.2641T>C (p.Phe881Leu)	ACGCTYTCAGGTGGCTGCCGG, CGCTYTCAGGTGGCTGCCGGGG	Hypercalciuric hypercalcemia
104893747	NM_198159.2(MITF):c.1195T>C (p.Ser399Pro)	ACTTYCCTTATTCCATCCACGG, CTTYCCCTTATTCCATCCACGGG	Waardenburg syndrome type 2A
104893770	NM_000539.3(RHO):c.133T>C (p.Phe45Leu)	CATGTTCTGCTGATCGTGTGG, ATGTTCTGCTGATCGTGTGGG	Retinitis pigmentosa 4
28937596	NM_003907.2(EIF2B5):c.1882T>C (p.Trp628Arg)	AGGCCYGGAGCCCTGTTTTAGG	Leukoencephalopathy with vanishing white matter
104893876	NM_001151.3(SLC25A4):c.293T>C (p.Leu98Pro)	GCAGCYCTCTAGGGGGTGTGG	Autosomal dominant progressive external ophthalmoplegia with mitochondrial DNA deletions 2
104893883	NM_006005.3(WFS1):c.2486T>C (p.Leu829Pro)	ACCATCCYGGAGGGCCGCTGGG	WFS1-Related Disorders
104893962	NM_000165.4(GJA1):c.52T>C (p.Ser18Pro)	CTACYCAACTGCTGGAGGGAAAGG	Oculodentodigital dysplasia
104893978	NM_000434.3(NEU1):c.718T>C (p.Trp240Arg)	GCCTCCYGGCGCTACGGAAGTGG, CCTCCYGGCGCTACGGAAGTGGG, CTCCYGGCGCTACGGAAGTGGGG	Sialidosis, type II
104894092	NM_002546.3(TNFRSF11B):c.349T>C (p.Phe117Leu)	TAGAGYCTGCTTGGAAACATAGG	Hyperphosphatasemia with bone disease
104894135	NM_000102.3(CYP17A1):c.316T>C (p.Ser106Pro)	CATCGCGYCCAACAAACCGTAAGG, ATCGCGYCCAACAAACCGTAAGGG	Complete combined 17-alpha-hydroxylase/17,20-lyase deficiency
104894151	NM_000102.3(CYP17A1):c.1358T>C (p.Phe453Ser)	AGCTCTYCCCTCATCATGGCCTGG	Combined partial 17-alpha-hydroxylase/17,20-lyase deficiency
36015961	NM_000518.4(HBB):c.344T>C (p.Leu115Pro)	TGTGTGCGGGCCCATCACTTTGG	Beta thalassemia intermedia
104894472	NM_152443.2(RDH12):c.523T>C (p.Ser175Pro)	TCCYCGGTGGCTACCACATTGG	Leber congenital amaurosis 13
104894587	NM_004870.3(MPDU1):c.356T>C (p.Leu119Pro)	TTCCYGGTCATGCACTACAGAGG	Congenital disorder of glycosylation type 1F
104894588	NM_004870.3(MPDU1):c.2T>C (p.Met1Thr)	AATAYGGCGGCCAGGGCGACGG	Congenital disorder of glycosylation type 1F
104894626	NM_000304.3(PMP22):c.82T>C (p.Trp28Arg)	TAGCAAYGGATCGTGGCAATGG	Charcot-Marie-Tooth disease, type IE
104894631	NM_018129.3(PNPO):c.784T>C (p.Ter262Gln)	ACCTYAACCTGGGACCTGCTGG	"Pyridoxal 5-phosphate-dependent epilepsy"
104894703	NM_032551.4(KISS1R):c.305T>C (p.Leu102Pro)	GCCCTGCGTACCCGCTGCCGGCTGG	
104894826	NM_000166.5(GJB1):c.407T>C (p.Val136Ala)	ATGTCATCAGCGTGGTGTCCGG	Dejerine-Sottas disease, X-linked hereditary motor and sensory neuropathy

104894859	NM_001122606.1(LAMP2):c.961T>C (p.Trp321Arg)	CAGCTACYGGGATGCCCGCCCTGG, AGCTACYGGGATGCCCGCCCTGGG	Danon disease
104894931	NM_006517.4(SLC16A2):c.1313T>C (p.Leu438Pro)	TGAGCYGGTGGGCCCAATGCAGG	Allan-Herndon-Dudley syndrome
104894935	NM_000330.3(RS1):c.38T>C (p.Leu13Pro)	TTACTTCYCTTGGCTATGAAGG	Juvenile retinoschisis
104895217	NM_001065.3(TNFRSF1A):c.175T>C (p.Cys59Arg)	TGCYGTACCAAGTGCCACAAAGG	TNF receptor-associated periodic fever syndrome (TRAPS)
143889283	NM_003793.3(CTSF):c.692A>G (p.Tyr231Cys)	CTCCAYACTGAGCTGTGCCACGG	Ceroid lipofuscinosis, neuronal, 13
122459147	NM_001159702.2(FHL1):c.310T>C (p.Cys104Arg)	GGGGYGCTTCAGGCCATTGTGG	Myopathy, reducing body, X-linked, childhood-onset
74552543	NM_020184.3(CNNM4):c.971T>C (p.Leu324Pro)	AAGCTCCYGGACTTTCTGGG	Cone-rod dystrophy amelogenesis imperfecta
199476117	m.10158T>C	AAAYCCACCCCTTACGAGTCGGG	Leigh disease, Leigh syndrome due to mitochondrial complex I deficiency, Mitochondrial complex I deficiency
794727808	NM_020451.2(SEPN1):c.872+2T>C	TTCCGGYGAGTGGGCCACACTGG	Congenital myopathy with fiber type disproportion, Eichsfeld type congenital muscular dystrophy
140547520	NM_005022.3(PFN1):c.350A>G (p.Glu117Gly)	CACCTYCTTGCCCCATCAGCAGG	Amyotrophic lateral sclerosis 18
397514359	NM_000060.3(BTD):c.445T>C (p.Phe149Leu)	TCACCGCYTCATGACACAGAGG	Biotinidase deficiency
207460001	m.15197T>C	CTAYCCGCCATCCCATACTTGG	Exercise intolerance
397514406	NM_000060.3(BTD):c.1214T>C (p.Leu405Pro)	TTCACCCYGGTCCCTGTCTGGGG	Biotinidase deficiency
397514516	NM_006177.3(NRL):c.287T>C (p.Met96Thr)	GAGGCCAYGGAGCTGTGCAGGG	Retinitis pigmentosa 27
72554312	NM_000531.5(OTC):c.134T>C (p.Leu45Pro)	CTCACTCYAAAAAACTTACCGG	Ornithine carbamoyltransferase deficiency
397514569	NM_178012.4(TUBB2B):c.350T>C (p.Leu117Pro)	GGTCCYGGATGTGGAGGAAGG	Polymicrogyria, asymmetric
397514571	NM_000431.3(MVK):c.122T>C (p.Leu41Pro)	CGGCYTCAACCCCCACAGCAATGG, GGCYTCAACCCCCACAGCAATGGG	Porokeratosis, disseminated superficial actinic 1
794728390	NM_000238.3(KCNH2):c.2396T>C (p.Leu799Pro)	GCCATCCYGGTATGGGGTGGGG, CCATCCYGGTATGGGGTGGGG, CATCCYGGTATGGGGTGGGGGG	Cardiac arrhythmia
397514713	NM_001199107.1(TBC1D24):c.686T>C (p.Phe229Ser)	GGTCTYTGACGTCTTCTGGTGG	Early infantile epileptic encephalopathy 16
397514719	NM_080605.3(B3GALT6):c.193A>G (p.Ser65Gly)	CGCYGGCCACCAGCACTGCCAGG	Spondyloepimetaphyseal dysplasia with joint laxity
730880608	NM_000256.3(MYBPC3):c.3796T>C (p.Cys1266Arg)	GAGYGCCGCCTGGAGGTGCGAGG	Cardiomyopathy
397515329	NM_001382.3(DPAGT1):c.503T>C (p.Leu168Pro)	AATCCYGTACTATGTCTACATGG, ATCCYGTACTATGTCTACATGGG, TCCYGTACTATGTCTACATGGGG	Congenital disorder of glycosylation type 1J
397515465	NM_018127.6(ELAC2):c.460T>C (p.Phe154Leu)	ATAYTTCTGGTCATTGAAAGG	Combined oxidative phosphorylation deficiency 17
397515557	NM_005211.3(CSF1R):c.2483T>C (p.Phe828Ser)	CATCTYTGACTGTGCTACACGG	Hereditary diffuse leukoencephalopathy with spheroids
397515599	NM_194248.2(OTOF):c.3413T>C (p.Leu1138Pro)	AGGTGCGYGTCTGGGGCTACGG, GGTGCYGTCTGGGGCTACGGG	Deafness, autosomal recessive 9
397515766	NM_000138.4(FBN1):c.2341T>C (p.Cys781Arg)	GGACAAYGTAGAAATACTCCTGG	Marfan syndrome
565779970	NM_001429.3(EP300):c.3573T>A (p.Tyr1191Ter)	CTTAYTACAGTTACAGAACAGG	Rubinstein-Taybi syndrome 2
786200938	NM_080605.3(B3GALT6):c.1A>G (p.Met1Val)	AGCTTCAYGGCGCCCGCGCCGGG, TCAYGGCGCCCGCGCCGGGGCG	Spondyloepimetaphyseal dysplasia with joint laxity
28942087	NM_000229.1(LCAT):c.698T>C (p.Leu233Pro)	ATCTCTYTGCTGGCTCCCTGGGG, TCTCYTGGGCTCCCTGGGGTGG	Norum disease
128621203	NM_000061.2(BTK):c.1625T>C (p.Leu542Pro)	TCGGCCYGTCCAGGTGAGTTGG	X-linked agammaglobulinemia with growth hormone deficiency
397515412	NM_006383.3(CIB2):c.368T>C (p.Ile123Thr)	CTTCAYCTGCAAGGAGGACCTGG	Deafness, autosomal recessive 48
193929364	NM_000352.4(ABCC8):c.404T>C (p.Leu135Pro)	AAGCYGCTAATTGGTAGGTGAGG	Permanent neonatal diabetes mellitus
730880872	NM_000257.3(MYH7):c.1400T>C (p.Ile467Thr)	TCGAGAYCTTCGATGTGAGTTGG, CGAGAYCTTCGATGTGAGTTGGG	Cardiomyopathy
80356474	NM_002977.3(SCN9A):c.2543T>C (p.Ile848Thr)	AAGATCAYTGGTAACTCAGTAGGG, AGATCAYTGGTAACTCAGTAGGG, GATCAYTGGTAACTCAGTAGGGGG	Primary erythromelalgia
80356489	NM_001164277.1(SLC37A4):c.352T>C (p.Trp118Arg)	GGGCYGGCCCCCATGTGGGAAGG	Glucose-6-phosphate transport defect
80356536	NM_152296.4(ATP1A3):c.2338T>C (p.Phe780Leu)	GCCCCYTCTGCTGTTCATCATGG	Dystonia 12
80356596	NM_194248.2(OTOF):c.3032T>C (p.Leu1011Pro)	GATGCGYGGTGGTCGACAACCTGG	Deafness, autosomal recessive 9, Auditory neuropathy, autosomal recessive, 1

80356689	NM_000083.2(CLCN1):c.857T>C (p.Val286Ala)	AGGAGYGCTATTTAGCATCGAGG	Myotonia congenita
118203884	m.4409T>C	AGGYCAGCTAAATAAGCTATCGG	Mitochondrial myopathy
587777625	NM_173596.2(SLC39A5):c.911T>C (p.Met304Thr)	AGAACAYGCTGGGCTTTGCAGG	Myopia 24, autosomal dominant
587783087	NM_003159.2(CDKL5):c.602T>C (p.Leu201Pro)	ATTCTYGGGGAGCTTAGCGATGG	not provided
118203951	NM_013319.2(UBIAD1):c.511T>C (p.Ser171Pro)	TCTGGCYCCTTCTCACACAGG, GGCYCCCTTCTACACAGGAGG	Schnyder crystalline corneal dystrophy
118204017	NM_000018.3(ACADVL):c.1372T>C (p.Phe458Leu)	TCGCATCYTCCGGATCTTGAGGG, GCATCYTCCGGATCTTGAGGGG	Very long chain acyl-CoA dehydrogenase deficiency
397518466	NM_000833.4(GRIN2A):c.2T>C (p.Met1Thr)	CTAYGGGCAGAGTGGGCTATTGG	Focal epilepsy with speech disorder with or without mental retardation
118204069	NM_000237.2(LPL):c.337T>C (p.Trp113Arg)	GGACYGGCTGTCACGGGCTCAGG	Hyperlipoproteinemia, type I
118204080	NM_000237.2(LPL):c.755T>C (p.Ile252Thr)	GTGAYTGCAGAGAGAGGACTTGG	Hyperlipoproteinemia, type I
118204111	NM_000190.3(HMBS):c.739T>C (p.Cys247Arg)	GCTTCGCCYGCATCGCTGAAAGGG	Acute intermittent porphyria
80357438	NM_007294.3(BRCA1):c.65T>C (p.Leu22Ser)	AAATCTYAGAGTGTCCCATCTGG	Familial cancer of breast, Breast-ovarian cancer, familial 1, Hereditary cancer-predisposing syndrome
139877390	NM_001040431.2(COA3):c.215A>G (p.Tyr72Cys)	CCAYCTGGGGAGGTAGGTTCAAGG	
793888527	NM_005859.4(PURA):c.563T>C (p.Ile188Thr)	GACCAYTGCCTGCCCGCGCAGGG, ACCAYTGCCTGCCCGCGCAGGG, CCAYTGCCTGCCCGCGCAGGGG	not provided, Mental retardation, autosomal dominant 31
561425038	NM_002878.3(RAD51D):c.1A>G (p.Met1Val)	CGCCCCAYGTTCCCCCGCAGGCCGG	Hereditary cancer-predisposing syndrome
121907934	NM_024105.3(ALG12):c.473T>C (p.Leu158Pro)	TCCYGCTGGCCCTCCCGGGCCTGG	Congenital disorder of glycosylation type 1G
80358207	NM_153212.2(GJB4):c.409T>C (p.Phe137Leu)	CCTCATCYTCAAGGCCGCCGTGG	Erythrokeratoderma variabilis
80358228	NM_002353.2(TACSTD2):c.557T>C (p.Leu186Pro)	TCGGCYGCACCCCAAGTCGTGG	Lattice corneal dystrophy Type III
121908076	NM_138691.2(TMC1):c.1543T>C (p.Cys515Arg)	AGGACCTYGCTGGGAAACAATGG, ACCTYGCTGGGAAACATGGTGG, CCTYGCTGGGAAACAATGGTGGG	Deafness, autosomal recessive 7
121908089	NM_017838.3(NHP2):c.415T>C (p.Tyr139His)	GGAGGCTYACGATGAGTGCCTGG, GGCTYACGATGAGTGCCTGGAGG	Dyskeratosis congenita autosomal recessive 1, Dyskeratosis congenita, autosomal recessive 2
121908154	NM_001243133.1(NLRP3):c.926T>C (p.Phe309Ser)	GGTGCCTYTGACGAGCACATAGG	Familial cold urticaria, Chronic infantile neurological, cutaneous and articular syndrome
121908158	NM_001033855.2(DCLRE1C):c.2T>C (p.Met1Thr)	GGCGCTAYGAGTTCTTCGAGGG, GCGCTAYGAGTTCTTCGAGGGGG	Histiocytic medullary reticulosis
796052870	NM_018129.3(PNPO):c.2T>C (p.Met1Thr)	CCCCCAYGACGTGCTGGCTGCAGGG, CCCAYGACGTGCTGGCTGCAGGGG	not provided
121908318	NM_020427.2(SLURP1):c.43T>C (p.Trp15Arg)	GCAGCCYGGAGCATGGCTGTGG	Acroerythrokeratoderma
121908352	NM_022124.5(CDH23):c.5663T>C (p.Phe1888Ser)	CTCACCTYCAACATCACTGCAGGG	Deafness, autosomal recessive 12
121908520	NM_000030.2(AGXT):c.613T>C (p.Ser205Pro)	CCTGTACYCGGGCTCCAGAAGG	Primary hyperoxaluria, type I
121908618	NM_004273.4(CHST3):c.920T>C (p.Leu307Pro)	CGTGCYGGCCTCGCCATGGTGG	Spondyloepiphyseal dysplasia with congenital joint dislocations
11694	NM_006432.3(NPC2):c.199T>C (p.Ser67Pro)	TATTCAGYCTAAAGCAGCAAGG	Niemann-Pick disease type C2
121908739	NM_000022.2(ADA):c.320T>C (p.Leu107Pro)	CCTGCYGGCCAACCTCCAAAGTGG	Severe combined immunodeficiency due to ADA deficiency
80359022	NM_000059.3(BRCA2):c.7958T>C (p.Leu2653Pro)	TGCYCTTCAACTAAAATACAGG	Familial cancer of breast, Breast-ovarian cancer, familial 2
121908902	NM_003880.3(WISP3):c.232T>C (p.Cys78Arg)	AAAATCYGTGCCAACGCAACCAGG, AAATCYGTGCCAACGCAACCAGGG, AATCYGTGCCAACGCAACCAGGG	Progressive pseudorheumatoid dysplasia
121908947	NM_006892.3(DNMT3B):c.808T>C (p.Ser270Pro)	CAAGTTCYCCGAGGTGAGTCCGG, AAGTCYCCGAGGTGAGTCCGGG, AGTTCYCCGAGGTGAGTCCGGGG	Centromeric instability of chromosomes 1,9 and 16 and immunodeficiency
121909028	NM_000492.3(CFTR):c.3857T>C (p.Phe1286Ser)	AGCCTYGGAGTGATACCAACAGG	Cystic fibrosis
121909135	NM_000085.4(CLCNKB):c.1294T>C (p.Tyr432His)	CTTTGTCYATGGTGAGTCTGGGG	Bartter syndrome type 3
121909143	NM_001300.5(KLF6):c.506T>C (p.Leu169Pro)	GGAGCYGCCCTCGCCAGGGAGG	
121909182	NM_001089.2(ABCA3):c.302T>C (p.Leu101Pro)	GCACYTGTGATCACATGCGAGG	Surfactant metabolism dysfunction, pulmonary, 3
121909200	NM_000503.5(EYA1):c.1459T>C (p.Ser487Pro)	CACTCYCGCTCATTCACTCCGG	Melnick-Fraser syndrome

121909247	NM_004970.2(IGFALS):c.1618T>C (p.Cys540Arg)	GGACYGTGGCTGCCCTCTCAAGG	Acid-labile subunit deficiency
121909253	NM_005570.3(LMAN1):c.2T>C (p.Met1Thr)	AGAYGGCGGGATCCAGGCAAAGG	Combined deficiency of factor V and factor VIII, 1
121909385	NM_000339.2(SLC12A3):c.1868T>C (p.Leu623Pro)	CAACCYGGCCCTCAGCTACTCGG	Familial hypokalemia-hypomagnesemia
121909497	NM_002427.3(MMP13):c.224T>C (p.Phe75Ser)	TTCTYCGGCTTAGAGGTGACTGG	Spondyloepimetaphyseal dysplasia, Missouri type
121909508	NM_000751.2(CHRND):c.188T>C (p.Leu63Pro)	AACCYCCTCTCCCTGGTGAGAGG	MYASTHENIC SYNDROME, CONGENITAL, 3B, FAST-CHANNEL
121909519	NM_001100.3(ACTA1):c.287T>C (p.Leu96Pro)	CGAGCYTCGCGTGGCTCCGAGG	Nemaline myopathy 3
121909572	NM_000488.3(SERPINC1):c.667T>C (p.Ser223Pro)	TGGGTGYCCAATAAGACCGAAGG	Antithrombin III deficiency
121909677	NM_000821.6(GGCX):c.896T>C (p.Phe299Ser)	TATGTYCTCCTACGTACGCTGG	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency
121909727	NM_001018077.1(NR3C1):c.2209T>C (p.Phe737Leu)	CTATTGCYTCCAAACATTGG	Glucocorticoid resistance, generalized
139573311	NM_000492.3(CFTR):c.1400T>C (p.Leu467Pro)	TTCACYCTAATGGTATTG, TCACYCTAATGGTATTG	Cystic fibrosis
121912441	NM_000454.4(SOD1):c.341T>C (p.Ile114Thr)	CATCAYTGGCCGCACACTGGTGG	Amyotrophic lateral sclerosis type 1
121912446	NM_000454.4(SOD1):c.434T>C (p.Leu145Ser)	CGTYYGGCTTGTTGTAATTGG, GTTYGGCTTGTTGTAATTGG	Amyotrophic lateral sclerosis type 1
121912463	NM_000213.3(ITGB4):c.1684T>C (p.Cys562Arg)	GGCCAGYGTGTGTGAGCCTGG	Epidermolysis bullosa with pyloric atresia
121912492	NM_002292.3(LAMB2):c.961T>C (p.Cys321Arg)	CCTCAACYGCAGCAGTCAGG	Nephrotic syndrome, type 5, with or without ocular abnormalities
397516659	NM_001399.4(EDA):c.2T>C (p.Met1Thr)	GGCCAYGGCTACCCGGAGGTGG	Hypohidrotic X-linked ectodermal dysplasia
111033589	NM_021044.2(DHH):c.485T>C (p.Leu162Pro)	GTTGCYGGCGCGCCTCGCAGTGG	46,XY gonadal dysgenesis, complete, dhh-related
111033622	NM_000206.2(IL2RG):c.343T>C (p.Cys115Arg)	TGGCYGTCAGTTGCAAAAAAAGG	X-linked severe combined immunodeficiency
121912613	NM_001041.3(SI):c.1859T>C (p.Leu620Pro)	ATGCYGGAGTTCAGTTGTTGG	Sucrase-isomaltase deficiency
121912619	NM_016180.4(SLC45A2):c.1082T>C (p.Leu361Pro)	GAGTTTCYCATCTACGAAAGAGG	Oculocutaneous albinism type 4
61750581	NM_000552.3(VWF):c.4837T>C (p.Ser1613Pro)	CTGCCYCTGATGAGATCAAGAGG	von Willebrand disease, type 2a
121912653	NM_000546.5(TP53):c.755T>C (p.Leu252Pro)	CATCCYACCATCATCACACTGG	Li-Fraumeni syndrome 1
111033683	NM_000155.3(GALT):c.386T>C (p.Met129Thr)	AGGTCACTGCTTCCACCCCTGG	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
111033752	NM_000155.3(GALT):c.677T>C (p.Leu226Pro)	CAGGAGCYACTCAGGAAGGTGG	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
121912729	NM_000039.1(APOA1):c.593T>C (p.Leu198Ser)	GCGCTYGGCCGCAGCCTTGAGG	Familial visceral amyloidosis, Ostertag type
769452	NM_000041.3(APOE):c.137T>C (p.Leu46Pro)	AACYGGCACTGGTCGCTTTGG	
121912762	NM_016124.4(RHD):c.329T>C (p.Leu110Pro)	ACACYGTTCAAGGTATTGGATGG	
111033824	NM_000155.3(GALT):c.1138T>C (p.Ter380Arg)	CGCCYGACCACGCCGACACAGG, GCCYGACCACGCCGACACAGG	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
111033832	NM_000155.3(GALT):c.980T>C (p.Leu327Pro)	TCCYGCGCTCTGCCACTGTCCGG	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
730881974	NM_000455.4(STK11):c.545T>C (p.Leu182Pro)	GGGAACCYGCTGCTACCACCGG, AACCYGCTGCTCACCACCGGTGG	Hereditary cancer-predisposing syndrome
1064644	NM_000157.3(GBA):c.703T>C (p.Ser235Pro)	GGGYCACTCAAGGGACAGCCCCGG	Gaucher disease
796052090	NM_138413.3(HOGA1):c.533T>C (p.Leu178Pro)	GGACCYGCCTGTGGATGCGAGTGG	Primary hyperoxaluria, type III
121913141	NM_000208.2(INSR):c.779T>C (p.Leu260Pro)	CTACCYGGACGGCAGGTGTGG	Leprechaunism syndrome
121913272	NM_006218.2(PIK3CA):c.1258T>C (p.Cys420Arg)	GGAACACYGTCCATTGGCATGGG, GAACACYGTCCATTGGCATGGGG	Congenital lipomatous overgrowth, vascular malformations, and epidermal nevi, Neoplasm of ovary, PIK3CA Related Overgrowth Spectrum
61751310	NM_000552.3(VWF):c.8317T>C (p.Cys2773Arg)	GCTCCYGCTGCTCCGACACGG	von Willebrand disease, type 2a
312262799	NM_024408.3(NOTCH2):c.1438T>C (p.Cys480Arg)	TTCACAYGTCTGTGCATGCCAGG	Alagille syndrome 2
121913570	NM_000426.3(LAMA2):c.7691T>C (p.Leu2564Pro)	ATCATTCTGGGAAGTGGAGGG, TCATTCYTTGGGAAGTGGAGGG	Merosin deficient congenital muscular dystrophy
121913640	NM_000257.3(MYH7):c.1046T>C (p.Met349Thr)	AACTCCAYGTATAAGCTGACAGG	Familial hypertrophic cardiomyopathy 1, Cardiomyopathy
121913642	NM_000257.3(MYH7):c.1594T>C (p.Ser532Pro)	CATCATGYCCATCCTGGAGAGG	Dilated cardiomyopathy 1S

119463996	NM_001079802.1(FKTN):c.527T>C (p.Phe176Ser)	GTAGTCTYTCATGAGAGGGAGTGG	Limb-girdle muscular dystrophy- dystroglycanopathy, type C4
587776456	NM_002049.3(GATA1):c.1240T>C (p.Ter414Arg)	GCTCAYGAGGGCACAGAGCATGG	GATA-1-related thrombocytopenia with dyserythropoiesis
63750654	NM_000184.2(HBG2):c.-228T>C	ATGCAAAATCTGTCTGAAACGG	Fetal hemoglobin quantitative trait locus 1
587776519	NM_001999.3(FBN2):c.3725-15A>G	AGCAYTGCAACCACATTGTCAGG	Congenital contractual arachnodactyly
78365220	NM_000402.4(G6PD):c.473T>C (p.Leu158Pro)	TGCCCYCCACCTGGGTACAGG	Anemia, nonspherocytic hemolytic, due to G6PD deficiency
63750741	NM_000179.2(MSH6):c.1346T>C (p.Leu449Pro)	CTGGGGCYGGTATTCTATGAAAGG	Hereditary Nonpolyposis Colorectal Neoplasms
587776914	NM_017565.3(FAM20A):c.590-2A>G	GTAATCYGCAAAGGAGGAGAAGG, TAATCYGCAAAGGAGGAGAAGGG	Enamel-renal syndrome
5030809	NM_000551.3(VHL):c.292T>C (p.Tyr98His)	CCCYACCCAACGCTGCCGCTGG	Von Hippel-Lindau syndrome, Hereditary cancer-predisposing syndrome
199476132	m.5728T>C	CAATCYACTTCTCCCGCCGCCGG, AATCYACTTCTCCGCCGCCGGG	Cytochrome-c oxidase deficiency, Mitochondrial complex I deficiency
62637012	NM_014336.4(AIPL1):c.715T>C (p.Cys239Arg)	CTGCCAGYGCCCTGCTGAAGAAGG, CCAGYGCCCTGCTGAAGAAGGAGG	Leber congenital amaurosis 4
199476199	NM_207352.3(CYP4V2):c.1021T>C (p.Ser341Pro)	AAACTGGYCCTTATACCTGTTGG, AACTGGYCCTTATACCTGTTGGG	Bietti crystalline corneoretinal dystrophy
587777183	NM_006702.4(PNPLA6):c.3053T>C (p.Phe1018Ser)	CCTYTAACCGCAGCATCCATCGG	Boucher Neuhauser syndrome
199476389	NM_000487.5(ARSA):c.899T>C (p.Leu300Ser)	GGTCTCTYGCAGGTGGAAAGGG	Metachromatic leukodystrophy
199476398	NM_016599.4(MYOZ2):c.142T>C (p.Ser48Pro)	TTAYCCCATCTCAGTAACCGTGG	Familial hypertrophic cardiomyopathy 16
119456967	NM_001037633.1(SIL1):c.1370T>C (p.Leu457Pro)	TTGCYGAAGGAGCTGAGATGAGG	Marinesco-Sjögren syndrome
730882253	NM_006888.4(CALM1):c.268T>C (p.Phe90Leu)	GGCAYTCAGGTCTTGACAAGG	Long QT syndrome 14
587777283	NM_012338.3(TSPAN12):c.413A>G (p.Tyr138Cys)	TAATCCAYAATTTCATCCTGG	Exudative vitreoretinopathy 5
587777306	NM_015884.3(MBTPS2):c.1391T>C (p.Phe464Ser)	GCTYTGCTTGATGGACAATGG	Palmarplantar keratoderma, mutilating, with periorificial keratotic plaques, X-linked
56378716	NM_000250.1(MPO):c.752T>C (p.Met251Thr)	TCACTCAYGTTCATGCAATGGGG	Myeloperoxidase deficiency
587777390	NM_005026.3(PIK3CD):c.1246T>C (p.Cys416Arg)	GCAGGACYGCCCCATTGCTGGG	Activated PI3K-delta syndrome
587777480	NM_003108.3(SOX11):c.178T>C (p.Ser60Pro)	TATGGYCCAAGATCGAACGCAAGG	Mental retardation, autosomal dominant 27
587777663	NM_001288767.1(ARMC5):c.1379T>C (p.Leu460Pro)	GCCCCGACYGCGGGATGCTGGTGG	Acth-independent macronodular adrenal hyperplasia 2
61753033	NM_000350.2(ABC4):c.5819T>C (p.Leu1940Pro)	AAGGCYACATGAACTAACCAAGG	Stargardt disease, Stargardt disease 1, Cone- rod dystrophy 3
200488568	NM_002972.3(SBF1):c.4768A>G (p.Thr1590Ala)	CAGGCGYCCTTGTCTCAGCCGG	Charcot-Marie-Tooth disease, type 4B3
132630274	NM_000377.2(WAS):c.809T>C (p.Leu270Pro)	CGGAGTCYGTTCAGGGCAGG	Severe congenital neutropenia X-linked
132630308	NM_001399.4(EDA):c.181T>C (p.Tyr61His)	CTGCYACCTAGAGTTGCGCTGG	Hypohidrotic X-linked ectodermal dysplasia
60934003	NM_170707.3(LMNA):c.1589T>C (p.Leu530Pro)	ACGGCTCYCATCAACTCCACTGG, CGGCTCYCATCAACTCCACTGGG, GGCTCYCATCAACTCCACTGGGG	Benign scapuloperoneal muscular dystrophy with cardiomyopathy
180177160	NM_000030.2(AGXT):c.1076T>C (p.Leu359Pro)	GGTGCYGCAGATGGCCCTGCTGG, GTGCYGCAGATGGCCCTGCTGGG	Primary hyperoxaluria, type I
180177222	NM_000030.2(AGXT):c.449T>C (p.Leu150Pro)	GTGCGYCTGTTCTAACCCACGG, TGCGYCTGTTCTAACCCACGGG	Primary hyperoxaluria, type I
180177254	NM_000030.2(AGXT):c.661T>C (p.Ser221Pro)	GCTCATCYCCTTCAGTGACAAGG	Primary hyperoxaluria, type I
180177264	NM_000030.2(AGXT):c.757T>C (p.Cys253Arg)	GGGGCYGTGACGACCAGCCAGG	Primary hyperoxaluria, type I
180177293	NM_000030.2(AGXT):c.893T>C (p.Leu298Pro)	GTATCYGCATGGGCCCTGCAGG	Primary hyperoxaluria, type I
376785840	NM_001282227.1(CECR1):c.1232A>G (p.Tyr411Cys)	GAAATCAYAGGACAAGCCTTGG	Polyarteritis nodosa
587779393	NM_000257.3(MYH7):c.4937T>C (p.Leu1646Pro)	GAGCCYCCAGAGCTTGTGAAGG	Myopathy, distal, 1
587779410	NM_012434.4(SLC17A5):c.500T>C (p.Leu167Pro)	ATTGTACYCAGAGCACTAGAAGG	Sialic acid storage disease, severe infantile type
587779513	NM_000090.3(COL3A1):c.2337+2T>C (p.Gly762_Lys779del)	AGGYAACCTTAATACTACCTGG	Ehlers-Danlos syndrome, type 4
777539013	NM_020376.3(PNPLA2):c.757+2T>C	GAACGGYGCAGGGACCCGGGGCGG, AACGGYGCAGGGACCCGGGGCGG	Neutral lipid storage disease with myopathy
34557412	NM_012452.2(TNFRSF13B):c.310T>C (p.Cys104Arg)	ACTTCYGTGAGAACAGCTCAGG	Immunoglobulin A deficiency 2, Common variable immunodeficiency 2
796052970	NM_001165963.1(SCN1A):c.1094T>C (p.Phe365Ser)	CAAGCTYTGATACCTCAGTTGG, AAGCTYTGATACCTCAGTTGG	not provided

724159989	NC_012920.1:m.7505T>C	CCTCCAYGACTTTTCAAAAAGG	Deafness, nonsyndromic sensorineural, mitochondrial
796053222	NM_014191.3(SCN8A):c.4889T>C (p.Leu1630Pro)	CGTCYGATCAAAGGCCAAAGGG, GTCYGATCAAAGGCCAAAGGG	not provided
118192127	NM_000540.2(RYR1):c.10817T>C (p.Leu3606Pro)	TACTACCYGGACCAGGTGGGTGG, ACTACCYGGACCAGGTGGGTGGG, CTACCYGGACCAGGTGGGTGGGG	Central core disease
118192170	NM_000540.2(RYR1):c.14693T>C (p.Ile4898Thr)	AGGCAYTGGGAGCAGATCGAGG	Malignant hyperthermia susceptibility type 1, Central core disease
121917703	NM_005247.2(FGF3):c.466T>C (p.Ser156Pro)	GTACGTGYCTGTGAACGGCAAGG, TACGTGYCTGTGAACGGCAAGGG	Deafness with labyrinthine aplasia microtia and microdontia (LAMM)
690016549	NM_005211.3(CSF1R):c.2450T>C (p.Leu817Pro)	CCGCCYGCCTGTGAATGGATGG	Hereditary diffuse leukoencephalopathy with spheroids
690016552	NM_005211.3(CSF1R):c.2566T>C (p.Tyr856His)	GAATCCCYACCCCTGGCATCCTGG	Hereditary diffuse leukoencephalopathy with spheroids
121917738	NM_001098668.2(SFTP42):c.593T>C (p.Phe198Ser)	GGAGACTYCCGCTACTCAGATGG, GAGACTYCCGCTACTCAGATGGG	Idiopathic fibrosing alveolitis, chronic form
690016559	NM_005211.3(CSF1R):c.1957T>C (p.Cys653Arg)	AGCCYGTACCCATGGAGGTAAGGG, GCCYGTACCCATGGAGGTAAGGG	Hereditary diffuse leukoencephalopathy with spheroids
690016560	NM_005211.3(CSF1R):c.2717T>C (p.Ile906Thr)	GCAGAYCTGCTCCTCTTCAGG	Hereditary diffuse leukoencephalopathy with spheroids
121917769	NM_003361.3(UMOD):c.376T>C (p.Cys126Arg)	GGCCACAYGTGTCATTGTGGTGG, GCCACAYGTGTCATTGTGGTGGG	Familial juvenile gout
121917773	NM_003361.3(UMOD):c.943T>C (p.Cys315Arg)	ATGGCACYGCCAGTGCAACAGG	Glomerulocystic kidney disease with hyperuricemia and isosthenuria
121917818	NM_007255.2(B4GALT7):c.617T>C (p.Leu206Pro)	TGCYCTCCAAGCAGCACTACCGG	Ehlers-Danlos syndrome progeroid type
121917824	NM_021615.4(CHST6):c.827T>C (p.Leu276Pro)	GGACCYGGCGCGGGAGCCGCTGG	Macular corneal dystrophy Type I
121917848	NM_000452.2(SLC10A2):c.728T>C (p.Leu243Pro)	TTTCYTCTGGCTAGAATTGCTGG	Bile acid malabsorption, primary
121918006	NM_000478.4(ALPL):c.1306T>C (p.Tyr436His)	TGGACYATGGTGAGAACCTCCAGG	Infantile hypophosphatasia
121918010	NM_000478.4(ALPL):c.979T>C (p.Phe327Leu)	CAAAGGCCYTCTTCTTGCTGGTGG, GGCYTCTTCTTGCTGGTGGAGG	Infantile hypophosphatasia
121918088	NM_000371.3(TTR):c.400T>C (p.Tyr134His)	CCCCYACT CCTATT CACCACGG	
121918110	NM_001042465.1(PSAP):c.1055T>C (p.Leu352Pro)	GAAGCYGCCGAAGTCCCCTGTCGG	Gaucher disease, atypical, due to saposin C deficiency
121918137	NM_003730.4(RNASET2):c.550T>C (p.Cys184Arg)	CCAGYGCCCTCCACCAAGCCAGG	Leukoencephalopathy, cystic, without megalecephaly
121918191	NM_001127628.1(FBP1):c.581T>C (p.Phe194Ser)	GGAGTYCATTTGGTGGACAAGG	Fructose-biphosphatase deficiency
121918306	NM_006946.2(SPTBN2):c.758T>C (p.Leu253Pro)	ACCAAGCYGCTGGATCCCGAAGGTGG, AAGCYGCTGGATCCCGAAGGTGG, AGCYGCTGGATCCCGAAGGTGG	Spinocerebellar ataxia 5
121918505	NM_000141.4(FGFR2):c.799T>C (p.Ser267Pro)	AATGCCYCCACAGTGGTCGGAGG	Pfeiffer syndrome, Neoplasm of stomach
121918643	NM_003126.2(SPTA1):c.620T>C (p.Leu207Pro)	GTGGAGCYGGTAGCTAAAGAAGG, TGGAGCYGGTAGCTAAAGAAGGG	Hereditary pyropoikilocytosis, Elliptocytosis 2
121918646	NM_001024858.2(SPTB):c.604T>C (p.Trp202Arg)	CTCCAGCYGGAAGGATGGCTGG	Spherocytosis type 2
121918648	NM_001024858.2(SPTB):c.6055T>C (p.Ser2019Pro)	ATGCCYCTGTGGCTGAGGGCTGG	
727504166	NM_000543.4(SMPD1):c.475T>C (p.Cys159Arg)	TGAGGCCYGTGGCTGCTCCTGG, GAGGCCYGTGGCTGCTCCTGGG	Niemann-Pick disease, type A, Niemann-Pick disease, type B
193922915	NM_000434.3(NEU1):c.1088T>C (p.Leu363Pro)	CAGCYATGGCCAGGCCCCAGTGG	Sialidosis, type II
727504419	NM_000501.3(ELN):c.889+2T>C	CAGGYAACATCTGTCAGCAGGG, AGGYAACATCTGTCAGCAGGG	Supravalvar aortic stenosis
376395543	NM_000256.3(MYBPC3):c.26-2A>G	GAGACYGAAGGGCCAGGTGGAGG	Primary familial hypertrophic cardiomyopathy, Familial hypertrophic cardiomyopathy 4, Cardiomyopathy
1169305	NM_000545.6(HNF1A):c.1720G>A (p.Gly574Ser)	GATGCGGCCAGGGTCTGGCTGG, ATGCGGCCAGGGTCTGGCTGGG, TGCGGCCAGGGTCTGGCTGGGG	Maturity-onset diabetes of the young, type 3
730880130	NM_000527.4(LDLR):c.1468T>C (p.Trp490Arg)	CTACYGGACCGACTCTGTCCTGG, TACYGGACCGACTCTGTCCTGGG	Familial hypercholesterolemia
281860286	NM_018713.2(SLC30A10):c.500T>C (p.Phe167Ser)	GGCGCTTYCGGGGGCCCTCAGGG	Hypermanganesemia with dystonia, polycythemia and cirrhosis
730880306	NM_145693.2(LPIN1):c.1441+2T>C	AAGGYACCGCGGGCTCGCGCGG, AGGYACCGCGGGCTCGCGCGGG	Myoglobinuria, acute recurrent, autosomal recessive
74315452	NM_000454.4(SOD1):c.338T>C (p.Ile113Thr)	TTGCAYCATTGGCCGCACACTGG	Amyotrophic lateral sclerosis type 1
730880455	NM_000169.2(GLA):c.41T>C (p.Leu14Pro)	CGCGCYTGCCTCGCTTCTGG	not provided
267606656	NM_054027.4(ANKH):c.1015T>C (p.Cys339Arg)	AGCTCYGTTCTGTGATGTTTGG	Craniometaphyseal dysplasia, autosomal dominant

267606687	NM_033409.3(SLC52A3):c.1238T>C (p.Val413Ala)	AGTTACGYCAAGGTGATGCTGGG	Brown-Vialetto-Van laere syndrome
267606721	NM_001928.2(CFD):c.640T>C (p.Cys214Arg)	GGTGYGCGGGGGCGTGCCTGAGGG, GTGYGCGGGGGCGTGCCTGAGGG	Complement factor d deficiency
267606747	NM_001849.3(COL6A2):c.2329T>C (p.Cys777Arg)	CGCCYCGACAAGCCACAGCAGG	Ullrich congenital muscular dystrophy
431905515	NM_001044.4(SLC6A3):c.671T>C (p.Leu224Pro)	CTGCACCCYCCACCAGAGCCATGG	Infantile Parkinsonism-dystonia
267606857	NM_000180.3(GUCY2D):c.2846T>C (p.Ile949Thr)	AGAGAYCGCCAACATGTCACTGG	Cone-rod dystrophy 6
267606880	NM_022489.3(INF2):c.125T>C (p.Leu42Pro)	GCTGCYCCAGATGCCCTGTGG	Focal segmental glomerulosclerosis 5
515726191	NM_015713.4(RRM2B):c.581A>G (p.Glu194Gly)	AACTCTYCTACAGCAGCAAAGG	RRM2B-related mitochondrial disease
267606917	NM_004646.3(NPHS1):c.793T>C (p.Cys265Arg)	GCTGCCYGCGTGGCCCCGAGGGGG, CTGCCYGCGTGGCCCCGAGGGGG	Finnish congenital nephrotic syndrome
267607104	NM_001199107.1(TBC1D24):c.751T>C (p.Phe251Leu)	CAAGTTCYCTCCACAAGGTGAGGG, TTCYTCCACAAGGTGAGGGCCGG	Myoclonic epilepsy, familial infantile
267607182	NM_144631.5(ZNF513):c.1015T>C (p.Cys339Arg)	TGGGCGCYGCATGCCAGGAGAGG, CGCYGCATGCGAGGAGAGGCTGG	Retinitis pigmentosa 58
267607211	NM_000229.1(LCAT):c.508T>C (p.Trp170Arg)	TATGACYGGCGGCTGGAGGCCGG	Norum disease
267607215	NM_016269.4(LEF1):c.181T>C (p.Ser61Pro)	GAACGAGYCTGAAATCATCCGG	Sebaceous tumors, somatic
587783580	NM_178151.2(DCX):c.683T>C (p.Leu228Pro)	AAAAAACYCTACACTCTGGATGG	Heterotopia
587783644	NM_004004.5(GJB2):c.107T>C (p.Leu36Pro)	GATCCYCGTTGTGGCTGCAAAGG	Hearing impairment
587783653	NM_005682.6(ADGRG1):c.1460T>C (p.Leu487Pro)	CCCTGCYCACCTGCCCTTCCTGG	Polymicrogyria, bilateral frontoparietal
587783863	NM_000252.2(MTM1):c.958T>C (p.Ser320Pro)	GGAACTTAAAAAAAGTGAAGG	Severe X-linked myotubular myopathy
267607751	NM_000249.3(MLH1):c.453+2T>C	ATCACGGYAAGAATGGTACATGG, TCACGGYAAGAATGGTACATGGG	Heredity Nonpolyposis Colorectal Neoplasms
119103227	NM_000411.6(HLCS):c.710T>C (p.Leu237Pro)	CTATCYTTCTCAGGGAGGGAGG	Holocarboxylase synthetase deficiency
119103237	NM_005787.5(ALG3):c.211T>C (p.Trp71Arg)	GATTGACYGGAAGGCCTACATGG	Congenital disorder of glycosylation type 1D
398122806	NM_003172.3(SURF1):c.679T>C (p.Trp227Arg)	CCACYGGCATTATCGAGACCTGG	Congenital myasthenic syndrome, acetazolamide-responsive
80338747	NM_004525.2(LRP2):c.7564T>C (p.Tyr2522His)	GTACCTGYACTGGCTGACTGGG	Donnai Barrow syndrome
398122838	NM_001271723.1(FBXO38):c.616T>C (p.Cys206Arg)	TTCCCTYGTATCCCAATGCTAAGG	Distal hereditary motor neuronopathy 2D
398122989	NM_014495.3(ANGPTL3):c.883T>C (p.Phe295Leu)	ACAAAACYTCAATGAAACGTGGG	Hypobetalipoproteinemia, familial, 2
80338945	NM_004004.5(GJB2):c.269T>C (p.Leu90Pro)	GCTCCYAGTGGCCATGCACGTGG	Deafness, autosomal recessive 1A, Hearing impairment
80338956	NM_000334.4(SCN4A):c.2078T>C (p.Ile693Thr)	AAGATCAYTGGCAATTCACTAGTGGGG, AGATCAYTGGCAATTCACTAGTGGGG, GATCAYTGGCAATTCACTAGTGGGG	Hyperkalemic Periodic Paralysis Type 1, Paramyotonia congenita of von Eulenburg
267608131	NM_000179.2(MSH6):c.4001+2T>C	CGGYAACTAACTAACTATAATGG	Heredity Nonpolyposis Colorectal Neoplasms
587784573	NM_004963.3(GUCY2C):c.2782T>C (p.Cys928Arg)	TCCCYGTGCTGCTGGAGTTGTGG, CCCYGTGCTGCTGGAGTTGTGG	Meconium ileus
267608511	NM_003159.2(CDKL5):c.659T>C (p.Leu220Pro)	CCAACYTTTACTATTCAAGAGG	Early infantile epileptic encephalopathy 2
373842615	NM_000118.3(ENG):c.1273-2A>G	CCGCCYCGGGGATAAAGCCAGG, CGCCYCGGGGATAAAGCCAGG	Haemorrhagic telangiectasia 1
185492581	NM_000335.4(SCN5A):c.376A>G (p.Lys126Glu)	GAATCTYCACAGCCCTCTCCGG	Brugada syndrome
200533370	NM_133499.2(SYN1):c.1699A>G (p.Thr567Ala)	GATGYCTGACGGGTAGCCTGTGG, ATGYCTGACGGGTAGCCTGTGG	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, not specified
118203981	NM_148960.2(CLDN19):c.269T>C (p.Leu90Pro)	GCTCCYGGGCTTCGTGGCCATGG	Hypomagnesemia 5, renal, with ocular involvement
137853892	NM_001235.3(SERPINH1):c.233T>C (p.Leu78Pro)	GTCGCYAGGGCTCGTCGCTGG, TCGCYAGGGCTCGTCGCTGG	Osteogenesis imperfecta type 10
118204024	NM_000263.3(NAGLU):c.142T>C (p.Phe48Leu)	GGCCGACYTCTCCGTGCGGTGG	Mucopolysaccharidosis, MPS-III-B
690016563	NM_005211.3(CSF1R):c.1745T>C (p.Leu582Pro)	CAACCYGCAGTTGGTGAGATGG	Heredity diffuse leukoencephalopathy with spheroids
58380626	NM_000526.4(KRT14):c.1243T>C (p.Tyr415His)	CGCCACCCYACCGCCGCTGCTGG, CACCYACCGCCGCTGCTGGAGG, ACCCYACCGCCGCCTGCTGGAGGG	Epidermolysis bullosa herpetiformis, Dowling-Meara
113994151	NM_207346.2(TSEN54):c.277T>C (p.Ser93Pro)	TTGAAGYCTCCCGCGGTGAGCGG, AAGYCTCCCGCGGTGAGCGGG	Pontocerebellar hypoplasia type 4
113994206	NM_004937.2(CTNS):c.473T>C (p.Leu158Pro)	TGGTCYAGGCTTCGACTTCGTGG	Cystinosis

62516109	NM_000277.1(PAH):c.638T>C (p.Leu213Pro)	CCACTTCYTGAAAAGTACTGTGG	Phenylketonuria
370011798	NM_001302946.1(TRNT1):c.668T>C (p.Ile223Thr)	GCAAYTCAGAAAATGCAAAGG	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay
62517167	NM_000277.1(PAH):c.293T>C (p.Leu98Ser)	AAGATCTYGAGGCATGACATTGG	Mild non-PKU hyperphenylalanemia
12021720	NM_001918.3(DBT):c.1150G>A (p.Gly384Ser)	GACYCACAGAGCCCAATTCTGG	Intermediate maple syrup urine disease type 2
104886289	NM_000495.4(COL4A5):c.4756T>C (p.Cys1586Arg)	TCCCCATYGTCTCAGGGATGGG	Alport syndrome, X-linked recessive
370471013	NC_012920.1:m.5559A>G	CAACYTACTGAGGGTTGAGG	Leigh disease
121434215	NM_000487.5(ARSA):c.410T>C (p.Leu137Pro)	GCCTTCCYGCCCCCCCATCAGGG	Metachromatic leukodystrophy, adult type
386134128	NM_000096.3(CP):c.1123T>C (p.Tyr375His)	ACACTACYACATTGCCGCTGAGG	Deficiency of ferroxidase
121434275	NM_001127328.2(ACADM):c.1136T>C (p.Ile379Thr)	GTGCAGAYACTTGAGGAATGG	Medium-chain acyl-coenzyme A dehydrogenase deficiency
121434276	NM_001127328.2(ACADM):c.742T>C (p.Cys248Arg)	CAGCGAYGTTCAGATACTAGAGG	Medium-chain acyl-coenzyme A dehydrogenase deficiency
121434284	NM_002225.3(IVD):c.134T>C (p.Leu45Pro)	ATGGGCYAAGCGAGGAGCAGAGG	IISOVALERIC ACIDEMIA, TYPE I
121434334	NM_005908.3(MANBA):c.1513T>C (p.Ser505Pro)	ATTACGYCCAGTCCTACAAATGG, TTACGYCCAGTCCTACAAATGGG, TACGYCCAGTCCTACAAATGGGG	Beta-D-mannosidosis
121434366	NM_000159.3(GCDH):c.883T>C (p.Tyr295His)	CGCCCGGYACGGCATCGCGTGGG, GCCCGGYACGGCATCGCGTGGGG	Glutaric aciduria, type 1
60715293	NM_000424.3(KRT5):c.541T>C (p.Ser181Pro)	GTTTGCCYCCTTCATCGACAAGG	Epidermolysis bullosa herpetiformis, Dowling-Meara
121434409	NM_001003722.1(GLE1):c.2051T>C (p.Ile684Thr)	AAGGACAYTCCTGTCCCCAAGGG	Lethal arthrogryposis with anterior horn cell disease
121434434	NM_001287.5(CLCN7):c.2297T>C (p.Leu766Pro)	GGGCCYCGGGCACCTGGTGGTGG	Osteopetrosis autosomal recessive 4
121434455	NM_000466.2(PEX1):c.1991T>C (p.Leu664Pro)	GATGACCYTGACCTCATTGCTGG	Zellweger syndrome
199422317	NM_001099274.1(TINF2):c.862T>C (p.Phe288Leu)	CTGYTCCCTTTAGGAATCTCGG	Aplastic anemia
104895221	NM_001065.3(TNFRSF1A):c.349T>C (p.Cys117Arg)	CTCTTCTYGCACAGTGGACCGGG	TNF receptor-associated periodic fever syndrome (TRAPS)
137854459	NM_000138.4(FBN1):c.4987T>C (p.Cys1663Arg)	GGGACAYGTTACAACACCCTGG	Marfan syndrome
387907075	NM_024027.4(COLEC11):c.505T>C (p.Ser169Pro)	CAGCTGYCCTGCCAGGGCGCGGG, AGCTGYCCTGCCAGGGCGCGGG, GCTGYCCTGCCAGGGCGCGGGGG, CTGYCCTGCCAGGGCGCGGGGG	Carnevale syndrome
1048095	NM_000352.4(ABCC8):c.674T>C (p.Leu225Pro)	TGCGYGTCCAAGGCACCTACTGG	Permanent neonatal diabetes mellitus
796065347	NM_019074.3(DLL4):c.1168T>C (p.Cys390Arg)	GAAYGTCCCCCCAACTTCACCGG	Adams-Oliver syndrome, ADAMS-OLIVER SYNDROME 6
137852347	NM_000402.4(G6PD):c.1054T>C (p.Tyr352His)	AGGGYACCTGGACGACCCCACGG	Anemia, nonspherocytic hemolytic, due to G6PD deficiency
74315327	NM_213653.3(HFE2):c.302T>C (p.Leu101Pro)	GGACCCYCGCCTTCCATTCCGGCG	Hemochromatosis type 2A
137852579	NM_000044.3(AR):c.2033T>C (p.Leu678Pro)	GTCCYGGAAGCCATTGAGCCAGG	
137852636	NM_001166107.1(HMGCS2):c.520T>C (p.Phe174Leu)	CCCTCYTCAATGCTGCCAACCTGG	mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency
137852661	NM_033163.3(FGF8):c.118T>C (p.Phe40Leu)	TTCCCTGYTCCGGCTGGCCGGG	Kallmann syndrome 6
121912967	NM_005215.3(DCC):c.503T>C (p.Met168Thr)	AGCCCAYGCCAACATCCACTGG	
137852806	NM_001039523.2(CHRNA1):c.901T>C (p.Phe301Leu)	TGTGYTCTCTGGTCATCGTGG	Myasthenic syndrome, congenital, fast-channel
137852850	NM_182760.3(SUMF1):c.463T>C (p.Ser155Pro)	GGCGACYCCTTGTCTTGAAGG	Multiple sulfatase deficiency
137852886	NM_000158.3(GBE1):c.671T>C (p.Leu224Pro)	AATGTACYACCAAGAACCAAAGG	Glycogen storage disease, type IV, GLYCOGEN STORAGE DISEASE IV, NONPROGRESSIVE HEPATIC
137852911	NM_000419.3(ITGA2B):c.641T>C (p.Leu214Pro)	CTGGTGCYTGGGGCTCTGGCGG	Glanzmann thrombasthenia
137852948	NM_138694.3(PKHD1):c.10658T>C (p.Ile3553Thr)	GAGCCCACTGAAATACGCTCAGG	Polycystic kidney disease, infantile type
137852964	NM_024960.4(PANK2):c.178T>C (p.Ser60Pro)	ATTGACYCAGTCGGATTCAATGG	
137853020	NM_006899.3(IDH3B):c.395T>C (p.Leu132Pro)	TGCGGCGYAGGGTAGGTGGCTGG, GCGGCGYAGGGTAGGTGGCTGGGG	Retinitis pigmentosa 46
137853249	NM_033500.2(HK1):c.1550T>C (p.Leu517Ser)	GAATCTYGGCCCTGGATCTTGG, TTCTYGGCCCTGGATCTTGGAGG	Hemolytic anemia due to hexokinase deficiency

137853270	NM_000444.5(PHEX):c.1664T>C (p.Leu555Pro)	AGCYCCAGAAGCCTTCAGGTGG	Familial X-linked hypophosphatemic vitamin D refractory rickets
137853325	NM_003639.4(IKBKG):c.1249T>C (p.Cys417Arg)	TGGAGYGCATTGAGTAGGGCCGG	Hypohidrotic ectodermal dysplasia with immune deficiency, Hyper-IgM immunodeficiency, X-linked, with hypohidrotic ectodermal dysplasia
28932769	NM_002055.4(GFAP):c.1055T>C (p.Leu352Pro)	GGACCGCTCAATGTCAGCTGG	Alexander disease
397507439	NM_002769.4(PRSS1):c.116T>C (p.Val39Ala)	TACCAGGYGTCCCTGAATTCTGG	Hereditary pancreatitis
387906446	NM_000132.3(F8):c.1729T>C (p.Ser577Pro)	AAAGAACTGTAGATCAAAGAGG	Hereditary factor VIII deficiency disease
387906482	NM_000133.3(F9):c.1031T>C (p.Ile344Thr)	ACGAACAYCTTCCTCAAATTGG	Hereditary factor IX deficiency disease
387906508	NM_000131.4(F7):c.983T>C (p.Phe328Ser)	GACGTYCTCTGAGAGGACGCTGG	Factor VII deficiency
387906532	NM_001040113.1(MYH11):c.3791T>C (p.Leu1264Pro)	GAAGCYGGAGGCGCAGGTGCAGG	Aortic aneurysm, familial thoracic 4
387906658	NM_002465.3(MYBPC1):c.2566T>C (p.Tyr856His)	CAAACCYATATCCGCAGAGTTGG	Distal arthrogryposis type 1B
387906701	NM_003491.3(NAA10):c.109T>C (p.Ser37Pro)	TGGCCTTYCCTGGCCCCAGGTGG, GGCCTTYCCTGGCCCCAGGTGGG	N-terminal acetyltransferase deficiency
387906717	NM_000377.2(WAS):c.881T>C (p.Ile294Thr)	GACTTCAYTGAGGACCCAGGGTGG, ACTTCACTGAGGACCCAGGGTGGG	Severe congenital neutropenia X-linked
387906809	NM_000287.3(PEX6):c.1601T>C (p.Leu534Pro)	CTTCYGGGCCGGGACCGTGATGG, TTCYGGGCCGGGACCGTGATGGG	Peroxisome biogenesis disorder 4B
387906965	NM_024513.3(FYCO1):c.4127T>C (p.Leu1376Pro)	CAGCCYGATCCCCATCACTGTGG	Cataract, autosomal recessive congenital 2
387906967	NM_006147.3(IRF6):c.65T>C (p.Leu22Pro)	GCCYCTACCCCTGGGCTCATCTGG	Van der Woude syndrome, Popliteal pterygium syndrome
387906982	NM_025132.3(WDR19):c.20T>C (p.Leu7Pro)	TCTCACYGCTAGAAAAGACTTGG	Asphyxiating thoracic dystrophy 5
387907072	NM_032446.2(MEGF10):c.2320T>C (p.Cys774Arg)	GGGCAGYGTACTTGGCCGACTGG	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant
137854499	NM_005502.3(ABCA1):c.6026T>C (p.Phe2009Ser)	GAGTYCTTGCCCTTTGAGAGG	Familial hypoalphalipoproteinemia
387907117	NM_000196.3(HSD11B2):c.1012T>C (p.Tyr338His)	CCGCCGCYATTACCCCGGCCAGG, CGCCGCYATTACCCCGGCCAGGG	Apparent mineralocorticoid excess
387907170	NM_004453.3(ETFDH):c.1130T>C (p.Leu377Pro)	CCAAACYCACCTTCTGGTGG	
387907205	NM_033360.3(KRAS):c.211T>C (p.Tyr71His)	GGACCAGYACATGAGGACTGGGG, CCAGYACATGAGGACTGGGGAGGG, CAGYACATGAGGACTGGGGAGGG	Cardiofaciocutaneous syndrome 2
387907240	NM_024110.4(CARD14):c.467T>C (p.Leu156Pro)	CAGCAGCYGCAGGAGCACCTGGG	Pityriasis rubra pilaris
387907282	NM_152296.4(ATP1A3):c.2431T>C (p.Ser811Pro)	TGCCATCYCACTGGCGTACGAGG	Alternating hemiplegia of childhood 2
387907361	NM_005120.2(MED12):c.3493T>C (p.Ser1165Pro)	AGGACYCTGAGCCAGGGGCCGG	Ohdo syndrome, X-linked
28933970	NM_006194.3(PAX9):c.62T>C (p.Leu21Pro)	GGCCGCGYGCCAACGCCATCCGG	Tooth agenesis, selective, 3
137854472	NM_000138.4(FBN1):c.3128A>G (p.Lys1043Arg)	TGCACAYTGCGTGGGTGCAGAGG	
727504261	NM_000257.3(MYH7):c.2708A>G (p.Glu903Gly)	AGCGCYCCTCAGCATGCCAGG	Cardiomyopathy, not specified
81002853	NM_000059.3(BRCA2):c.476-2A>G	ACCACYGGGGTAAAAAAAGGGG, TACCACYGGGGTAAAAAAAGGG, ATACCACYGGGGTAAAAAAAGG	Familial cancer of breast, Breast-ovarian cancer, familial 2, Hereditary cancer-predisposing syndrome
119473032	NM_021020.3(LZTS1):c.355A>G (p.Lys119Glu)	CCCTYCTCGGAGCCCTGTAGAGG	
193922801	NM_000540.2(RYR1):c.7043A>G (p.Glu2348Gly)	TTCYCCCTCACGCTCTGCCCTGG	not provided
36210419	NM_000218.2(KCNQ1):c.652A>G (p.Lys218Glu)	GCCCCTYGGAGCCACGCGAGG	Torsades de pointes, Cardiac arrhythmia
121964989	NM_000108.4(DLD):c.1483A>G (p.Arg495Gly)	TTCTCYAAAGCTCTGATAAGG	Maple syrup urine disease, type 3
28936669	NM_000095.2(COMP):c.1418A>G (p.Asp473Gly)	ATTGCGTCGTCGTCGCGCAGG	
28936696	NM_018488.2(TBX4):c.1592A>G (p.Gln531Arg)	GTACYGTAAGGAAGATTCTCGGG, GGTACYGTAAGGAAGATTCTCGG	Ischiopatellar dysplasia
121965077	NM_000137.2(FAH):c.1141A>G (p.Arg381Gly)	TCCYGGTCTGACCATTCCCCAGG	Tyrosinemia type I
794728203	NM_000138.4(FBN1):c.3344A>G (p.Asp1115Gly)	ACTCAYCAATATCTGAAATGG	Thoracic aortic aneurysms and aortic dissections
786205436	NM_003002.3(SDHD):c.275A>G (p.Asp92Gly)	GAATAGYCCATCGCAGAGCAAGG	Fatal infantile mitochondrial cardiomyopathy
72551317	NM_000784.3(CYP27A1):c.776A>G (p.Lys259Arg)	AGTCCACYTGGGGAGGAAGGTGG	Cholestanol storage disease

786205687	NM_016218.2(POLK):c.1385A>G (p.Asn462Ser)	ATTCACAYTCTTCAACTTAATGG	Malignant tumor of prostate
794728280	NM_000138.4(FBN1):c.7916A>G (p.Tyr2639Cys)	TGTTCACAYTCTTCAAGGGCGGG, CTGTTCACAYTCTTCAAGGGCGGG	Thoracic aortic aneurysms and aortic dissections
28937317	NM_000335.4(SCN5A):c.3971A>G (p.Asn1324Ser)	GCAYTGACCACCACCTCAAGTGG	Long QT syndrome 3, Congenital long QT syndrome
786205854	NM_144499.2(GNAT1):c.386A>G (p.Asp129Gly)	CGGAGYCCCTTCCACAGCCGCTGG	NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1G
104893776	NM_000539.3(RHO):c.533A>G (p.Tyr178Cys)	GGATGYACCTGAGGACAGGCCAGG	Retinitis pigmentosa 4
28937590	NM_001257342.1(BCS1L):c.232A>G (p.Ser78Gly)	GACACYGAGGTGCTGAGTACGGG, CGACACYGAGGTGCTGAGTACGG	GRACILE syndrome
104893866	NM_000320.2(QDPR):c.449A>G (p.Tyr150Cys)	TGCCGYACCCGATCATACCTGGG, ATGCCGYACCCGATCATACCTGG	Dihydropteridine reductase deficiency
587776590	NM_015629.3(PRPF31):c.527+3A>G	GACAYACCCCTGGGTGGTGGAGG, GCGGACAYACCCCTGGGTGGTGG	Retinitis pigmentosa 11
104894015	NM_000162.3(GCK):c.641A>G (p.Tyr214Cys)	GTAGYAGCAGGAGATCATCGTGG	Hyperinsulinemic hypoglycemia familial 3
202247823	NM_000532.4(PCCB):c.1606A>G (p.Asn536Asp)	ATATYTGCATGTTTCTCCAAGG	Propionic acidemia
104894199	NM_000073.2(CD3G):c.1A>G (p.Met1Val)	CCAYGTCAGTCTGTCCTCCGG	Immunodeficiency 17
104894208	NM_001814.4(CTSC):c.857A>G (p.Gln286Arg)	CTCCYGAGGGCTTAGGATTGGGG, CCTCCYGAGGGCTTAGGATTGGG, ACCTCCYGAGGGCTTAGGATTGG	Papillon-Lefèvre syndrome, Haim-Munk syndrome
104894211	NM_001814.4(CTSC):c.1040A>G (p.Tyr347Cys)	TCCTACAYAGTGGTACTCAGAGG	Papillon-Lefèvre syndrome, Periodontitis, aggressive, 1
104894290	NM_000448.2(RAG1):c.2735A>G (p.Tyr912Cys)	CTGYACTGGCAGAGGGATTCTGG	Histiocytic medullary reticulosis
104894354	NM_000217.2(KCNA1):c.676A>G (p.Thr226Ala)	GCGYTTCCACGATGAAGAAGGGG, AGCGYTTCCACGATGAAGAAGGG, CAGCGYTTCCACGATGAAGAAGG	Episodic ataxia type 1
104894425	NM_014239.3(EIF2B2):c.638A>G (p.Glu213Gly)	AGTTGTCYCAATACCTGCTTGG	Leukoencephalopathy with vanishing white matter, Ovarioleukodystrophy
104894450	NM_000270.3(PNP):c.383A>G (p.Asp128Gly)	ATAYCTCCAACCTCAAACCTGGG, GATAYCTCCAACCTCAAACCTGG	Purine-nucleoside phosphorylase deficiency
147394623	NM_024887.3(DHDDS):c.124A>G (p.Lys42Glu)	GGCACTYCTTGGCATAGCGACGG	Retinitis pigmentosa 59
60723330	NM_005557.3(KRT16):c.374A>G (p.Asn125Ser)	GCGGTCACTGAGGTTCTGCATGG	Pachyonychia congenita, type 1, Palmoplantar keratoderma, nonepidermolytic, focal
104894634	NM_030665.3(RAI1):c.4685A>G (p.Gln1562Arg)	CTGCTGCGTCTCGTCGCTGCGTTGG	Smith-Magenis syndrome
104894730	NM_000363.4(TNNI3):c.532A>G (p.Lys178Glu)	CCTYCTTACCTGCTTGAGGTGG, CCTCCTYCTTCACCTGCTTGAGG	Familial restrictive cardiomyopathy 1
104894816	NM_002049.3(GATA1):c.653A>G (p.Asp218Gly)	GTCCTGYCCCTCCGCCACAGTGG	GATA-1-related thrombocytopenia with dyserythropoiesis
794726773	NM_001165963.1(SCN1A):c.1662+3A>G	GTGCCAYACCTGGTGTGGGGAGG	Severe myoclonic epilepsy in infancy
104894861	NM_000202.6(IDS):c.404A>G (p.Lys135Arg)	AAAGACTYTTCCCACCGACATGG	Mucopolysaccharidosis, MPS-II
104894874	NM_000266.3(NDP):c.125A>G (p.His42Arg)	TGGYGCCTCATGCAGCGTCGAGG	
191205969	NM_002420.5(TRPM1):c.296T>C (p.Leu99Pro)	AAGCYCTTAATATCTGTGCATGG	Congenital stationary night blindness, type 1C
794727073	NM_019109.4(ALG1):c.1188-2A>G	TAAACYGCAAGAGAGAACCAAGGG, GTAAACYGCAAGAGAGAACCAAGG	Congenital disorder of glycosylation type 1K
281875236	NM_001004334.3(GPR179):c.659A>G (p.Tyr220Cys)	CCCACAYATCCATCTGCCTGCGG	Congenital stationary night blindness, type 1E
28939094	NM_015915.4(ATL1):c.1222A>G (p.Met408Val)	CACCCAYCTTCTCACCCCTCGG	Spastic paraparesis 3
281875324	NM_005359.5(SMAD4):c.989A>G (p.Glu330Gly)	ATCCATTYCAAAGTAAGCAATGG	Juvenile polyposis syndrome, Hereditary cancer-predisposing syndrome
77173848	NM_000037.3(ANK1):c.-108T>C	GGGCCYGGCCCGCACGTACAGG	Spherocytosis, type 1, autosomal recessive
150181226	NM_001159772.1(CANT1):c.671T>C (p.Leu224Pro)	CGTCYGTACGTGGCCGGCTGGG, GCGTCYGTACGTGGCCGGCTGG	Desbuquois syndrome
397514253	NM_000041.3(APOE):c.237-2A>G	CGCCCCYGGCCCGAGAGGGCGGG, GCGCCCYGGCCCGAGAGGGCGG	Familial type 3 hyperlipoproteinemia
397514348	NM_000060.3(BTD):c.278A>G (p.Tyr93Cys)	GTTCACTGCAAGGTTCTGG	Biotinidase deficiency
397514415	NM_000060.3(BTD):c.1313A>G (p.Tyr438Cys)	GGCAYACAGCTCTTGGATAAGG	Biotinidase deficiency
397514501	NM_007171.3(POMT1):c.430A>G (p.Asn144Asp)	GAGCATYCTCTGTTCAAAGAGG	Limb-girdle muscular dystrophy-dystroglycanopathy, type C1
370382601	NM_174917.4(ACSF3):c.1A>G (p.Met1Val)	GGCAGCAYTGCAGTACGACAGGCAG	not provided
72554332	NM_000531.5(OTC):c.238A>G (p.Lys80Glu)	AAGGACTYCCCTTGCAATAAGG	Ornithine carbamoyltransferase deficiency
397514599	NM_033109.4(PNPT1):c.1424A>G	GACTYCAGATGTAACCTTATGG	Deafness, autosomal recessive 70

	(p.Glu475Gly)		
397514650	NM_000108.4(DLD):c.1444A>G (p.Arg482Gly)	GACTCYAGCTATATCTTCACAGG	Maple syrup urine disease, type 3
397514675	NM_003156.3(STIM1):c.251A>G (p.Asp84Gly)	TTCCACAYCCACATACCATTGG	Myopathy with tubular aggregates
794728378	NM_000238.3(KCNH2):c.1913A>G (p.Lys638Arg)	ATCYTCTCTGAGTTGGTGTGTTGG, GATCYTCTCTGAGTTGGTGTGTTGG	Cardiac arrhythmia
397514711	NM_002163.2(IRF8):c.238A>G (p.Thr80Ala)	AACCTCGYCTTCCAAGTGGCTGG	Autosomal dominant CD11C+/CD1C+ dendritic cell deficiency
397514729	NM_000388.3(CASR):c.85A>G (p.Lys29Glu)	CCCCCTYCTTTGGGCTCGCTGG	Hypocalcemia, autosomal dominant 1, with bartter syndrome
397514743	NM_022114.3(PRD16):c.2447A>G (p.Asn816Ser)	GCCGCCGYTTGGCTGGCACGGG	Left ventricular noncompaction 8
397514757	NM_005689.2(ABCB6):c.508A>G (p.Ser170Gly)	TGGGCYGTTCAGAACACCAGGG, GTGGGCYGTTCAGAACACCAGGG	Dyschromatosis universalis hereditaria 3
28940313	NM_152443.2(RDH12):c.677A>G (p.Tyr226Cys)	CACTGCGYAGGTGGTACCCGG	Leber congenital amaurosis 13
794728538	NM_000218.2(KCNQ1):c.1787A>G (p.Glu596Gly)	GTCTYCTACTCGGTTCAAGGCAGGG, TGTCYCTACTCGGTTCAAGGCAGGG	Cardiac arrhythmia
794728569	NM_000218.2(KCNQ1):c.605A>G (p.Asp202Gly)	AGGYCTGTGGAGTGCAGGAGAGG	Cardiac arrhythmia
794728573	NM_000218.2(KCNQ1):c.1515-2A>G	GCCYGCAGTGGAGAGAGGAGAGG	Cardiac arrhythmia
370874727	NM_003494.3(DYSF):c.3349-2A>G	CCGCCCYGGAGACACGAAGCTGG	Limb-girdle muscular dystrophy, type 2B
794728859	NM_198056.2(SCN5A):c.2788-2A>G	ACCYGTCGAGATAATGGTCAGG	not provided
794728887	NM_198056.2(SCN5A):c.4462A>G (p.Thr1488Ala)	CCTCTGTCATGAAGATGTCTGG	not provided
28940878	NM_000372.4(TYR):c.125A>G (p.Asp42Gly)	CTCCTGYCCCCGCTCCACGGTGG	Tyrosinase-negative oculocutaneous albinism
397515420	NM_172107.2(KCNQ2):c.1636A>G (p.Met546Val)	GCAYGACACTGCAGGGGGGTGGG, CGCAYGACACTGCAGGGGGGTGG, AACCGCAYGACACTGCAGGGGG	Early infantile epileptic encephalopathy 7
397515428	NM_001410.2(MEGF8):c.7099A>G (p.Ser2367Gly)	GACYCCCGTGAAATGATCCCGG	Carpenter syndrome 2
143601447	NM_201631.3(TGM5):c.122T>C (p.Leu41Pro)	TCAACCYCACCCTGTACTTCAGG	Peeling skin syndrome, acral type
397515519	NM_000207.2(INS):c.*59A>G	GGGCYTTATTCCATCTCTCTCGG	Permanent neonatal diabetes mellitus
397515523	NM_000370.3(TTPA):c.191A>G (p.Asp64Gly)	CAGGYCCAGATCGAAATCCCGGG, CCAGGYCCAGATCGAAATCCCGGG	Ataxia with vitamin E deficiency
397515891	NM_000256.3(MYBPC3):c.1224-2A>G	TACTTGCYGTAGAACAGAAAGGG	Familial hypertrophic cardiomyopathy 4, Cardiomyopathy
397516082	NM_000256.3(MYBPC3):c.927-2A>G	GTCCCYGTGTCCCGCAGTCAGG	Familial hypertrophic cardiomyopathy 4, Cardiomyopathy
397516138	NM_000257.3(MYH7):c.2206A>G (p.Ile736Val)	TATCAAYGAACCTGCCTCCAGGG, CTATCAAYGAACCTGCCTCCAGGG	Familial hypertrophic cardiomyopathy 1, Cardiomyopathy, not specified
1154510	NM_002150.2(HPD):c.97G>A (p.Ala33Thr)	ATGACGYGGCCTGAATCACAGGG, AATGACGYGGCCTGAATCACAGGG	4-Alpha-hydroxyphenylpyruvate hydroxylase deficiency
397516330	NM_000260.3(MYO7A):c.6439-2A>G	ATATCCYGGGGAGCAGAAAGGG, GATATCCYGGGGAGCAGAAAGGG	Usher syndrome, type 1
72556271	NM_000531.5(OTC):c.482A>G (p.Asn161Ser)	CAGCCCAYTGATAATTGGGATGG	not provided
606231260	NM_023073.3(C5orf42):c.3290-2A>G	ATCYATCAAATACAAAAATTGG	Orofaciodigital syndrome 6
587777521	NM_004817.3(TJP2):c.1992-2A>G	CAGCTCYGAGAAGAAACCACGGG, TCAGCTCYGAGAAGAAACCACGG	Progressive familial intrahepatic cholestasis 4
730880846	NM_000257.3(MYH7):c.617A>G (p.Lys206Arg)	CTTCYTGCTGCGGTCCCCAATGG	Cardiomyopathy
397517978	NM_206933.2(USH2A):c.12067-2A>G	TTCCCYGTAAGAAAATTAACAGG	Usher syndrome, type 2A, Retinitis pigmentosa 39
606231409	NM_000216.2(ANOS1):c.1A>G (p.Met1Val)	GCACCACTGGCTGCGGGTCGAGGG, GGCACCACTGGCTGCGGGTCGAGGG	Kallmann syndrome 1
80356546	NM_003334.3(UBA1):c.1639A>G (p.Ser547Gly)	TGGCYTGTCAACCCGGATATGTGG	Arthrogryposis multiplex congenita, distal, X-linked
80356584	NM_194248.2(OTOF):c.766-2A>G	GACCYGCAGGCAGGGAGAAGGGGG, TGACCYGCAGGCAGGGAGAAGGGGG, CTGACCYGCAGGCAGGGAGAAGGG, GCTGACCCYGCAGGCAGGGAGAAGGG	Deafness, autosomal recessive 9
730880930	NM_000257.3(MYH7):c.1615A>G (p.Met539Val)	GGAACAYGCACCTCCTTCCAGG	Cardiomyopathy
118203947	NM_013319.2(UBIAD1):c.355A>G (p.Arg119Gly)	TCCYGTCACTCTTTGTGG	Schnyder crystalline corneal dystrophy
60171927	NM_000526.4(KRT14):c.368A>G (p.Asn123Ser)	GCGGTCACTGAGGTTCTGCATGG	Epidermolysis bullosa herpetiformis, Dowling-Meara
199422248	NM_001363.4(DKC1):c.941A>G (p.Lys314Arg)	AATCYGGCCCCATAGCAGATGG	Dyskeratosis congenita X-linked

72558467	NM_000531.5(OTC):c.929A>G (p.Glu310Gly)	TCCACTYCTTCTGGCTTCTGGG, ATCCACTYCTTCTGGCTTCTGG	not provided
72558478	NM_000531.5(OTC):c.988A>G (p.Arg330Gly)	ACTTCYGTGTTCTGCCTCTGG, CACTTCYGTGTTCTGCCTCTGG	not provided
118204455	NM_000505.3(F12):c.158A>G (p.Tyr53Cys)	GGTGGYACTGGAAGGGAAAGTGG	
80357477	NM_007294.3(BRCA1):c.5453A>G (p.Asp1818Gly)	TTGYCCTCTGTCCAGGCATCTGG	Familial cancer of breast, Breast-ovarian cancer, familial 1
121907908	NM_024426.4(WT1):c.1021A>G (p.Ser341Gly)	CGCYCTCGTACCCCTGCTGTGG	Mesothelioma
121907926	NM_000280.4(PAX6):c.1171A>G (p.Thr391Ala)	GTGGYGCCCGAGGTGCCATTGG	Optic nerve aplasia, bilateral
121908023	NM_024740.2(ALG9):c.860A>G (p.Tyr287Cys)	TTAYACAAAACAATGTTGAGTGG	Congenital disorder of glycosylation type 1L
121908148	NM_001243133.1(NLRP3):c.1880A>G (p.Glu627Gly)	ACAATYCCAGCTGGCTGGGCTGG	Familial cold urticaria
121908166	NM_006492.2(ALX3):c.608A>G (p.Asn203Ser)	CGGYTCTGGAACCAGACCTGGG, GCGGYTCTGGAACCAGACCTGG, TCGGYTCTGGAACCAGACCTGG	Frontonasal dysplasia 1
121908184	NM_020451.2(SEPN1):c.1A>G (p.Met1Val)	CCCAYGGCTCGGGCTGGCGGG, CGGCCCCAYGGCTGGCGTGGCG	Eichsfeld type congenital muscular dystrophy
121908258	NM_130468.3(CHST14):c.878A>G (p.Tyr293Cys)	AAGTCAYAGTGCACGGCACAAAGG	Ehlers-Danlos syndrome, musculocontractural type
121908383	NM_001128425.1(MUTYH):c.1241A>G (p.Gln414Arg)	AAGCYGCTCTGAGGGCTCCAGG	Neoplasm of stomach
121908580	NM_004328.4(BCS1L):c.148A>G (p.Thr50Ala)	GTGYGATCATGTAATGGCGCCGG	Mitochondrial complex III deficiency
121908584	NM_016417.2(GLRX5):c.294A>G (p.Gln98=)	CCTGACCYTGTGGAGCTCCGGG	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive
121908635	NM_022817.2(PER2):c.1984A>G (p.Ser662Gly)	GCCACACYCTCTGCCTTGCCGG	Advanced sleep phase syndrome, familial
121908655	NM_003839.3(TNFRSF11A):c.508A>G (p.Arg170Gly)	GGGTCYGCATTTGTCCGTGGAGG	Osteopetrosis autosomal recessive 7
29001653	NM_000539.3(RHO):c.886A>G (p.Lys296Glu)	CGCTCTYGGCAAAGAACGCTGG, GCGCTCTYGGCAAAGAACGCTGG	Retinitis pigmentosa 4
56307355	NM_006502.2(POLH):c.1603A>G (p.Lys535Glu)	AGACTTTYCTGCTAAAGAAGGG	Xeroderma pigmentosum, variant type
121908919	NM_002977.3(SCN9A):c.1964A>G (p.Lys655Arg)	CCTTTCTYGTGTATTGATTGG	Generalized epilepsy with febrile seizures plus, type 7, not specified
121908939	NM_006892.3(DNMT3B):c.2450A>G (p.Asp817Gly)	GACACGYCTGTGTAGTGCACAGG	Centromeric instability of chromosomes 1,9 and 16 and immunodeficiency
121909088	NM_001005360.2(DNM2):c.1684A>G (p.Lys562Glu)	ACTYCTCTCTTCTCCTGAGGG, TACTYCTCTCTTCTCCTGAGG	Charcot-Marie-Tooth disease, dominant intermediate b, with neutropenia
120074112	NM_000483.4(APOC2):c.1A>G (p.Met1Val)	GCCCCAYAGTGTCCAGAGACCTGG	Apolipoprotein C2 deficiency
121909239	NM_000314.6(PTEN):c.755A>G (p.Asp252Gly)	ATAYCACCAACACACAGGTAACGG	Macrocephaly/autism syndrome
121909251	NM_198217.2(ING1):c.515A>G (p.Asn172Ser)	TGGYTGCACAGACAGTACGTGGG, CTGGYTGCACAGACAGTACGTGG	Squamous cell carcinoma of the head and neck
121909396	NM_001174089.1(SLC4A11):c.2518A>G (p.Met840Val)	GATCAYCTTCATGTAGGGCAGGG, AGATCAYCTTCATGTAGGGCAGG	Corneal dystrophy and perceptive deafness
121909533	NM_000034.3(ALDOA):c.386A>G (p.Asp129Gly)	CCAYCCAACCTAAAGAGAACAGG	HNSHA due to aldolase A deficiency
128627255	NM_004006.2(DMD):c.835A>G (p.Thr279Ala)	TGACCGYGATCTGCAGAGAACGG, CTGACCGYGATCTGCAGAGAACGG	Dilated cardiomyopathy 3B
116929575	NM_001085.4(SERPINA3):c.1240A>G (p.Met414Val)	GCTCAYGAAGAAGATGTTCTGGG, TGCTCAYGAAGAAGATGTTCTGG	
61748392	NM_004992.3(MECP2):c.410A>G (p.Glu137Gly)	CAACYCCACTTCTAGAGCGAAAGG	Mental retardation, X-linked, syndromic 13
61748906	NM_001005741.2(GBA):c.667T>C (p.Trp223Arg)	CCCACTYGGCTCAAGACCAATGG	Gaucher disease, type 1
199473024	NM_000238.3(KCNH2):c.3118A>G (p.Ser1040Gly)	CTGCYCTCCACGTCGCCCCGGGG, CCTGCYCTCCACGTCGCCCCGGGG, GCCTGCYCTCCACGTCGCCCCGG	Sudden infant death syndrome
794728365	NM_000238.3(KCNH2):c.1129-2A>G	GGACCYGCACCCGGGGAGGCAGG	Cardiac arrhythmia
72556293	NM_000531.5(OTC):c.548A>G (p.Tyr183Cys)	AGAGCTAYAGTGTCTCTAAAGG	not provided
111033244	NM_000441.1(SLC26A4):c.1151A>G (p.Glu384Gly)	TGAATYCTTAAGGAAGAGACTGG	Pendred syndrome, Enlarged vestibular aqueduct syndrome
111033415	NM_000260.3(MYO7A):c.1344-2A>G	AGCYGCAGGGGCACAGGGATGGG, AAGCYGCAGGGGCACAGGGATGG	Usher syndrome, type 1
121912439	NM_000454.4(SOD1):c.302A>G (p.Glu101Gly)	AGAATCTYCAATAGACACATCGG	Amyotrophic lateral sclerosis type 1
111033567	NM_002769.4(PRSS1):c.68A>G (p.Lys23Arg)	ATCYTGTCAATCATCATCAAAGG, GATCYTGTCAATCATCATCAAAGG	Hereditary pancreatitis
121912565	NM_000901.4(NR3C2):c.2327A>G (p.Gln776Arg)	TCATCYGTTGCCTGCTAAGCGG	Pseudohypoaldosteronism type 1 autosomal dominant

121912574	NM_000901.4(NR3C2):c.2915A>G (p.Glu972Gly)	CCGACYCCACCTGGGCAGCTGG	Pseudohypoaldosteronism type 1 autosomal dominant
121912589	NM_001173464.1(KIF21A):c.2839A>G (p.Met947Val)	ATTCAYATCTGCCCTCATGTTGG	Fibrosis of extraocular muscles, congenital, 1
111033661	NM_000155.3(GALT):c.253-2A>G	ATTCACCYACCGACAAGGATAGG	Deficiency of UDPglucose-hexose-1-phosphate uridyltransferase
111033669	NM_000155.3(GALT):c.290A>G (p.Asn97Ser)	GAAGTCGYTGTCAACAGGAAGG	Deficiency of UDPglucose-hexose-1-phosphate uridyltransferase
111033682	NM_000155.3(GALT):c.379A>G (p.Lys127Glu)	TGACCTYACTGGGTGGTGACGGG, ATGACCTYACTGGGTGGTGACGG	Deficiency of UDPglucose-hexose-1-phosphate uridyltransferase
111033786	NM_000155.3(GALT):c.950A>G (p.Gln317Arg)	CAGCYGCCAATGGTTCAGTTGG	Deficiency of UDPglucose-hexose-1-phosphate uridyltransferase
121912765	NM_001202.3(BMP4):c.278A>G (p.Glu93Gly)	CCTCCYCCCCAGACTGAAGCCGG	Microphthalmia syndromic 6
121912856	NM_000094.3(COL7A1):c.425A>G (p.Lys142Arg)	CACCYTGGGACACCAAGTCGGG, TCACCYTGGGACACCAAGTCGG	Epidermolysis bullosa dystrophica inversa, autosomal recessive
199474715	NM_152263.3(TPM3):c.505A>G (p.Lys169Glu)	CCAACYTACGAGGCCACCTACAGG	Congenital myopathy with fiber type disproportion
199474718	NM_152263.3(TPM3):c.733A>G (p.Arg245Gly)	ATCYCTCAGCAAACCTCAGCACGG	Congenital myopathy with fiber type disproportion
121912895	NM_001844.4(COL2A1):c.2974A>G (p.Arg992Gly)	CCTCYCTCACCACTGGTGCAGG	Spondyloepimetaphyseal dysplasia Strudwick type
121913074	NM_000129.3(F13A1):c.851A>G (p.Tyr284Cys)	ATAGGCAYAGATAATTGTCAGG	Factor xiii, a subunit, deficiency of
121913145	NM_000208.2(INSR):c.707A>G (p.His236Arg)	GCTGYGGCAACAGAGGCCTTCGG	Leprechaunism syndrome
312262745	NM_025137.3(SPG11):c.2608A>G (p.Ile870Val)	ACTTAYCCTGGGGAGAAGGATGG	Spastic paraparesis 11, autosomal recessive
121913682	NM_000222.2(KIT):c.2459A>G (p.Asp820Gly)	AGAACYATTCTTGATGTCCTCTGG	Mast cell disease, systemic
587776757	NM_000151.3(G6PC):c.230+4A>G	GTTCYTACCACTTAAAGACGAGG	Glycogen storage disease type 1A
61752063	NM_000330.3(RS1):c.286T>C (p.Trp96Arg)	TTCTTCGYGGACTGCAAACAGG	Juvenile retinoschisis
367543065	NM_024549.5(TCTN1):c.221-2A>G	AGCAACYGCAAAAAAGAGGGG, CAGCAACYGCAAAAAAGAGGG	Joubert syndrome 13
5030773	NM_000894.2(LHB):c.221A>G (p.Gln74Arg)	CCACCYGAGGCAGGGCGGCAGG	Isolated lutropin deficiency
199476092	NM_000264.3(PTCH1):c.2479A>G (p.Ser827Gly)	CGTTACYGAAACTCCTGTGTAGG	Gorlin syndrome, Holoprosencephaly 7, not specified
398123158	NM_000117.2(EMD):c.450-2A>G	CGTCCCCYAGGGCAAAAGAGGGG	not provided
199476103	RMRP:n.71A>G	ACTTYCCCCTAGGCGGAAAGGGG, GACTTYCCCCTAGGCGGAAAGGG, GGACTTYCCCCTAGGCGGAAAGGG	Metaphyseal chondrodysplasia, McKusick type, Metaphyseal dysplasia without hypotrichosis
5030856	NM_000277.1(PAH):c.1169A>G (p.Glu390Gly)	CTCYCTGCCACGTAATACAGGGG, ACTCYCTGCCACGTAATACAGGG, AACTCYCTGCCACGTAATACAGG	Phenylketonuria, Hyperphenylalaninemia, non-pku
5030860	NM_000277.1(PAH):c.1241A>G (p.Tyr414Cys)	GGGTCGYAGCGAACTGAGAAGGG, TGGGTCGYAGCGAACTGAGAAGG	Phenylketonuria, Hyperphenylalaninemia, non-pku
587777055	NM_020988.2(GNAO1):c.521A>G (p.Asp174Gly)	GGATGYCCTGCTCGGTGGCTGG	Early infantile epileptic encephalopathy 17
587777223	NM_024301.4(FKRP):c.1A>G (p.Met1Val)	CCGCAYGGGCCGAAGTCTGGGG, GCCGCAYGGGCCGAAGTCTGGG, AGCCGCAYGGGCCGAAGTCTGG	Congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies type A5
587777479	NM_003108.3(SOX11):c.347A>G (p.Tyr116Cys)	GTACTTGAGTCGGGTAGTCGG	Mental retardation, autosomal dominant 27
587777496	NM_020435.3(GJC2):c.-170A>G	TTGYTCCCCCTCGGCCCTCAGGG, ATTGYTCCCCCTCGGCCCTCAGGG	Leukodystrophy, hypomyelinating, 2
587777507	NM_022552.4(DNMT3A):c.1943T>C (p.Leu648Pro)	CTCCYGGTCTGAAGGACTTGGG, GCTCCYGGTCTGAAGGACTTGG	Tatton-Brown-rahman syndrome
587777557	NM_018400.3(SCN3B):c.482T>C (p.Met161Thr)	AATCAYGATGTACATCCTCTGG	Atrial fibrillation, familial, 16
587777569	NM_001030001.2(RPS29):c.149T>C (p.Ile50Thr)	GATAYCGGTTTCATTAAGGTAGG	Diamond-Blackfan anemia 13
587777657	NM_153334.6(SCARF2):c.190T>C (p.Cys64Arg)	CCACGYGCTGCGCTGGCTGGAGG	Marden Walker like syndrome
587777689	NM_005726.5(TSFM):c.57+4A>G	ACTTCYCACCAGGGTAGCTCCCGG	Combined oxidative phosphorylation deficiency 3
796052005	NM_000255.3(MUT):c.329A>G (p.Tyr110Cys)	GCAYACTGGCGGATGGTCCAGGG, AGCAYACTGGCGGATGGTCCAGGG	not provided
587777809	NM_144596.3(TTC8):c.115-2A>G	GTTCCYGGAAAGCATTAAAGAAGG	Retinitis pigmentosa 51
587777878	NM_000166.5(GJB1):c.580A>G (p.Met194Val)	TAGCAYGAAGACGGTGAAGACGG	X-linked hereditary motor and sensory neuropathy
74315420	NM_001029871.3(RSPO4):c.194A>G (p.Gln65Arg)	CGTACYGGCGGATGCCCTCCGG	Anonychia
180177219	NM_000030.2(AGXT):c.424-2A>G (p.Gly_142Gln145del)	AGGCCCYGAGGAAGCAGGGACGG	Primary hyperoxaluria, type I

367610201	NM_002693.2(POLG):c.1808T>C (p.Met603Thr)	CTCAYGGCACTTACCTGGGATGG	not provided
180177319	NM_012203.1(GRHPR):c.84-2A>G	TCACAGCGGGGAAAGGGAGG	Primary hyperoxaluria, type II
796052068	NM_000030.2(AGXT):c.777-2A>G	GGTACCYGGAAGACACGAGGGG, TGGTACCYGGAAGACACGAGGGG	Primary hyperoxaluria, type I
61754010	NM_000552.3(VWF):c.1583A>G (p.Asn528Ser)	TGCCAYGTAAATTCCACACAGG	von Willebrand disease, type 2a
587778866	NM_000321.2(RB1):c.1927A>G (p.Lys643Glu)	ATTYCAATGGCTCTGGGCTGG	Retinoblastoma
74435397	NM_006331.7(EMG1):c.257A>G (p.Asp86Gly)	ATAYCTGGCCGCGCTCCCCAGG	Bowen-Conradi syndrome
796052527	NM_000156.5(GAMT):c.1A>G (p.Met1Val)	CGCTCAYGCTGCAGGCTGGACGG	not provided
796052637	NM_172107.2(KCNQ2):c.848A>G (p.Lys283Arg)	GTACYTGTCCCCGTAGCCAATGG	not provided
724159963	NM_032228.5(FAR1):c.1094A>G (p.Asp365Gly)	GATAYCATACAGGAATGCTGGGG, AGATAYCATACAGGAATGCTGGG, TAGATAYCATACAGGAATGCTGG	Peroxisomal fatty acyl-coa reductase 1 disorder
587779722	NM_000090.3(COL3A1):c.1762-2A>G (p.Gly588_Gln605del)	CACCCYAAAGAAGAAGTGGTCGG	Ehlers-Danlos syndrome, type 4
118192102	m.8296A>G	TTTACAGYGGGCTCTAGAGGGGG	Diabetes-deafness syndrome maternally transmitted
727502787	NM_001077494.3(NFKB2):c.2594A>G (p.Asp865Gly)	CTGYCTTCCTTCACCTCTGCTGG	Common variable immunodeficiency 10
727503036	NM_000117.2(EMD):c.266-2A>G	AGCCYTGGGAAGGGGGCAGCGG	Emery-Dreifuss muscular dystrophy 1, X-linked
690016544	NM_005861.3(STUB1):c.194A>G (p.Asn65Ser)	GGCCCCGGYTGGTGTAAACACGG	Spinocerebellar ataxia, autosomal recessive 16
690016554	NM_005211.3(CSF1R):c.2655-2A>G	GTATCYGGGAGATAGGACAGAGG	Hereditary diffuse leukoencephalopathy with spheroids
118192185	NM_172107.2(KCNQ2):c.1A>G (p.Met1Val)	GCACCAYGGTGCCTGGCGGGAGG	Benign familial neonatal seizures 1
121917869	NM_012064.3(MIP):c.401A>G (p.Glu134Gly)	AGATCYCCACTGTGGTTGCCTGG	Cataract 15, multiple types
121918014	NM_000478.4(ALPL):c.1250A>G (p.Asn417Ser)	AGGCCCAYTGCCATACAGGATGG	Infantile hypophosphatasia
121918036	NM_000174.4(GP9):c.110A>G (p.Asp37Gly)	GCAGYCCACCCACAGCCCCATGG	Bernard-Soulier syndrome type C
121918089	NM_000371.3(TTR):c.379A>G (p.Ile127Val)	CGGCAAYGGTGTAGCGGCGGGGG, GCGGCAAYGGTGTAGCGGCGGGGG	Amyloidogenic transthyretin amyloidosis
121918121	NM_000823.3(GHRHR):c.985A>G (p.Lys329Glu)	CGACTYGGAGAGACCCCTGCAGG	Isolated growth hormone deficiency type 1B
121918333	NM_015335.4(MED13L):c.6068A>G (p.Asp2023Gly)	ATATCAYCTAGAGGGAAAGGGGG, CATATCAYCTAGAGGGAAAGGGGG	Transposition of great arteries
121918605	NM_001035.2(RYR2):c.12602A>G (p.Gln4201Arg)	CGCCAGCYGCATTCAAAGATGG	Catecholaminergic polymorphic ventricular tachycardia
587781262	NM_002764.3(PRPS1):c.343A>G (p.Met115Val)	TAGCAYATTTGCAACAAGCTTGG	Charcot-Marie-Tooth disease, X-linked recessive, type 5, Deafness, high-frequency sensorineural, X-linked
121918608	NM_001161766.1(AHCY):c.344A>G (p.Tyr15Cys)	GCGGGYACTTGGTGTGGATGAGG	Hypermethioninemia with s-adenosylhomocysteine hydrolase deficiency
121918613	NM_000702.3(ATP1A2):c.1033A>G (p.Thr345Ala)	CTGYCAGGGTCAGGCACACCTGG	Familial hemiplegic migraine type 2
587781339	NM_000535.5(PMS2):c.904-2A>G	GCAGACCYGCACAAAATACAAGG	Hereditary cancer-predisposing syndrome
121918691	NM_001128177.1(THRB):c.1324A>G (p.Met442Val)	CTTCAYGTGCAGGAAGCGGCTGG	Thyroid hormone resistance, generalized, autosomal dominant
121918692	NM_001128177.1(THRB):c.1327A>G (p.Lys443Glu)	CCACCTYCATGTGCAGGAAGCGG	Thyroid hormone resistance, generalized, autosomal dominant
727504333	NM_000256.3(MYBPC3):c.2906-2A>G	CCGTTCYGTGGGTATAGAGTGGG, GCCGTTCYGTGGGTATAGAGTGG	Familial hypertrophic cardiomyopathy 4
786200910	NM_006204.3(PDE6C):c.1483-2A>G	CTTTCYGTGAAATAAGGATGGG, TCTTTCYGTGAAATAAGGATGG	Achromatopsia 5
281860296	NM_000551.3(VHL):c.586A>T (p.Lys196Ter)	GGTCTTYCTGCACATTGGGTGG	Von Hippel-Lindau syndrome
730880444	NM_000169.2(GLA):c.370-2A>G	GTGAACCYGAAATGAGAGGGAGG	not provided
730880531	NM_000256.3(MYBPC3):c.1227-2A>G	GTACCCYGGTGGGGCCGCAGGG, TGTACCCYGGTGGGGCCGCAGGG	Familial hypertrophic cardiomyopathy 4, Cardiomyopathy
267606643	NM_013411.4(AK2):c.494A>G (p.Asp165Gly)	TCAYCTTCATGGGCTCTTTGG	Reticular dysgenesis
267606705	NM_005188.3(CBL):c.1144A>G (p.Lys382Glu)	TATTTYACATAGTGGAAATGTGG	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia
62642934	NM_000277.1(PAH):c.916A>G (p.Ile306Val)	GGCCAAYTCCCTGTAAATTGGGG, AGGCCAAYTCCCTGTAAATTGGGG	Phenylketonuria, Hyperphenylalaninemia, non-pku
267606782	NM_000117.2(EMD):c.1A>G (p.Met1Val)	TCCAYGGCGGGTGCGGGCTCAGG	Emery-Dreifuss muscular dystrophy, X-linked
267606820	NM_014053.3(FLVCR1):c.361A>G (p.Asn121Asp)	AGGCCTYGACCAGCGAGTACAGG	Posterior column ataxia with retinitis pigmentosa

730880805	NM_000257.3(MYH7):c.4664A>G (p.Glu1555Gly)	GCCCCCTCGTGCTCCAGGGAGG, CTTGGCCCYCCTCGTGCTCCAGGG	Cardiomyopathy
267606834	NM_138387.3(G6PC3):c.346A>G (p.Met116Val)	TGATCAYGCAGTGTCCAGAAGGG, GTGATCAYGCAGTGTCCAGAAGGG	Dursun syndrome
267606851	NM_000175.3(GPI):c.1028A>G (p.Gln343Arg)	GTACYGGTCATAGGGCAGCATGG	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency
515726182	NM_015713.4(RRM2B):c.190T>C (p.Trp64Arg)	TTCCCTCYGGACAGCAGAAAGAGG	RRM2B-related mitochondrial disease
730881002	NM_002880.3(RAF1):c.1279A>G (p.Ser427Gly)	GCTGCGGCCCTCGCACCACTGGG, GGCTGCGGCCCTCGCACCACTGG	Rasopathy
267607030	NM_002977.3(SCN9A):c.29A>G (p.Gln10Arg)	AAGCTCYGAGGTCTGGGGGAGG	Primary erythromelalgia
267607048	NM_007373.3(SHOC2):c.4A>G (p.Ser2Gly)	TACYCAGGGTACTCAAGCCTGG	Noonan-like syndrome with loose anagen hair, Rasopathy
587783486	NM_004380.2(CREBBP):c.3983-2A>G	GCAGCCCYAGGAAGTCCAGAAGG	Rubinstein-Taybi syndrome
730881357	NM_000051.3(ATM):c.3154-2A>G	AGCCYACGGGAAAAGAACTGTGG	Hereditary cancer-predisposing syndrome
398122404	NM_001256864.1(DNAJC6):c.801-2A>G	AGGTATCYGAAACAGAAGGTTGG	Parkinson disease 19, juvenile-onset
267607482	NM_001927.3(DES):c.1024A>G (p.Asn342Asp)	GAATCGTYCTGCAGGAGAGGGGG	Myofibrillar myopathy 1
796053439	NM_000391.3(TPP1):c.833A>G (p.Gln278Arg)	CAGGTACYGCACATCTAGACTGG	not provided
587783835	NM_000252.2(MTM1):c.550A>G (p.Arg184Gly)	GTTATT CYCCAATGGTGATTGGG	Severe X-linked myotubular myopathy
587783842	NM_000252.2(MTM1):c.629A>G (p.Asp210Gly)	TCATCAYCTGAGGCACGATAACGG	Severe X-linked myotubular myopathy
267607777	NM_000249.3(MLH1):c.884+4A>G	TGCTACAYTACCTGAGGTACAGG	Hereditary Nonpolyposis Colorectal Neoplasms
33972047	NM_000518.4(HBB):c.59A>G (p.Asn20Ser)	CACGYTCACCTTGCCCCACAGGG, CCACGYTCACCTTGCCCCACAGG	alpha Thalassemia
730882004	NM_000546.5(TP53):c.709A>G (p.Met237Val)	ACACAYGTAGTTGTAGTGGATGG	Li-Fraumeni syndrome, Hereditary cancer-predisposing syndrome
730882052	NM_001231.4(CASQ1):c.731A>G (p.Asp244Gly)	GGCTTGYCTGGGATGGTCACAGG	Myopathy, vacuolar, with casq1 aggregates
80338959	NM_000334.4(SCN4A):c.4078A>G (p.Met1360Val)	GATCAYGATGGTGATGTCGAAGG	Hyperkalemic Periodic Paralysis Type 1
80338960	NM_000334.4(SCN4A):c.4108A>G (p.Met1370Val)	CCATCAYGGTGACCATGTTGAGG	Hyperkalemic Periodic Paralysis Type 1
80338962	NM_000334.4(SCN4A):c.4774A>G (p.Met1592Val)	TGTACAYGTTGACCACGATGAGG	Hyperkalemic Periodic Paralysis Type 1, Familial hyperkalemic periodic paralysis
398123062	NM_012160.4(FBXL4):c.1694A>G (p.Asp565Gly)	TATGYCCAGCTGCTGTAAACCTGG	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type)
730882140	NM_001039550.1(DNAJB2):c.14A>G (p.Tyr5Cys)	GATCTCGYAGTAGGATGCCATGG	Charcot-Marie-Tooth disease, Charcot-Marie-Tooth disease, axonal, type 2T
796053522	NM_052859.3(RFT1):c.1222A>G (p.Met408Val)	GCAYCACAAAATTGTACCTGGGG, AGCAYCACAAAATTGTACCTGGG, CAGCAYCACAAAATTGTACCTGG	Congenital disorder of glycosylation type 1N
398123211	NM_000169.2(GLA):c.548-2A>G	AACCYGTATGAGAAAACAATGGG, TAACCYGTATGAGAAAACAATGG	Fabry disease
587784423	NM_006306.3(SMC1A):c.616-2A>G	AGCCYGTGCAAACAGGGGAATGG	Congenital muscular hypertrophy-cerebral syndrome
398123411	NM_000487.5(ARSA):c.1108-2A>G	GGCTCYGGGGCAGAGTCAGGGG, GGGCTCYGGGGCAGAGTCAGGG, AGGGCTCYGGGGCAGAGTCAGG	Metachromatic leukodystrophy
398123429	NM_000512.4(GALNS):c.1171A>G (p.Met391Val)	CCGCCAYCAGCGTGTGCCACGG	Mucopolysaccharidosis, MPS-IV-A
267608500	NM_003159.2(CDKL5):c.578A>G (p.Asp193Gly)	ATGYCCACGGACTTCCATAGGG, CATGYCCACGGACTTCCATAGG	Early infantile epileptic encephalopathy 2
398123552	NM_000402.4(G6PD):c.188T>C (p.Ile63Thr)	ACACACAYATTCATCATCATGGG	Anemia, nonspherocytic hemolytic, due to G6PD deficiency
75391579	NM_000155.3(GALT):c.563A>G (p.Gln188Arg)	TTACCYGGCAGTGGGGTGGGGG, CTTACCYGGCAGTGGGGTGGGG, CCTTACCYGGCAGTGGGGTGGGG	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
398123639	NM_001848.2(COL6A1):c.805-2A>G	TTCTCCCYGGAACACAAAACAGG	Ullrich congenital muscular dystrophy, Bethlem myopathy
398123750	NM_003482.3(KMT2D):c.5645-2A>G	GCAGTTCYGTGGGGGAATGAAGG	Kabuki make-up syndrome
398124528	NM_144997.5(FLCN):c.1433-2A>G	CCCACYGGGAGAACGGGCAGGGG, GCCACYGGGAGAACGGGCAGGG, GGCCACYGGGAGAACGGGCAGG	Hereditary cancer-predisposing syndrome
113994149	NM_025265.3(TSEN2):c.926A>G (p.Tyr309Cys)	CAGAGCAYAGACCAAGAAAAAGG	Pontocerebellar hypoplasia type 2B
281865052	NM_198578.3(LRRK2):c.5605A>G (p.Met1869Val)	TCAACAYAATTTCTAGGCAGG	Parkinson disease 8, autosomal dominant
281865495	NM_004614.4(TK2):c.562A>G (p.Thr188Ala)	AAGYCTCAGGATTGGTCCGAAGG	Mitochondrial DNA depletion syndrome 2

756328339	NM_003494.3(DYSF):c.3041A>G (p.Tyr1014Cys)	CTAYACTCCCAGCCTGGGGAGG, ATGCTAYACTCCCAGCCTGGGG, GATGCTAYACTCCCAGCCTGGGG	Limb-girdle muscular dystrophy, type 2B
387906810	NM_153427.2(PITX2):c.262A>G (p.Lys88Glu)	TCTYGAACCAAACCTGGGGCGG, GATTCTYGAACCAAACCTGGGG, CGATTCTYGAACCAAACCTGGGG	Axenfeld-Rieger syndrome type 1
78310959	NM_030964.3(SPRY4):c.530A>G (p.Lys177Arg)	AGTCYCTGTCCAGCTCGGGTGG, AAGTCYCTGTCCAGCTCGGGTGG	Hypogonadotropic hypogonadism 17 with or without anosmia
144109267	NM_207352.3(CYP4V2):c.1393A>G (p.Arg465Gly)	TTCCYGGGGCCAGCAGAGAAGGG, GTTCCYGGGGCCAGCAGAGAAGGG	Bietti crystalline corneoretinal dystrophy
104886319	NM_000495.4(COL4A5):c.1340-2A>G	CACCYGAGTAAGATAAAAGAAAGG	Alport syndrome, X-linked recessive
104886416	NM_000495.4(COL4A5):c.466-2A>G	ACCCYAAAAGAACGCCATCAATGG	Alport syndrome, X-linked recessive
121434443	NM_004984.2(KIF5A):c.827A>G (p.Tyr276Cys)	GAACAYAGCTTTCTGGGGAGG	Spastic paraplegia 10
199422314	NM_001099274.1(TINF2):c.850A>G (p.Thr284Ala)	TGACTGYGGGGCGCTCCTTATGG	Dyskeratosis congenita autosomal dominant
121434478	NM_004044.6(ATIC):c.1277A>G (p.Lys426Arg)	AGTGTACYTGACAGCAATGGTGG	AICAR transformylase/IMP cyclohydrolase deficiency
111033765	NM_000155.3(GALT):c.812A>G (p.Glu271Gly)	CGCYCAGCAGGGTCAGCTCAGG	Deficiency of UDPglucose-hexose-1-phosphate uridyltransferase
121434606	NM_006006.4(ZBTB16):c.1849A>G (p.Met617Val)	GATCAYGGCCGACTAGTCCCCGG, TGATCAYGGCCGAGTAGTCCCCGG	Skeletal defects, genital hypoplasia, and mental retardation
566325901	NM_000017.3(ACADS):c.1108A>G (p.Met370Val)	AGCCCAYGCCGCCAGGATCTGG	not provided
148665132	NM_012079.5(DGAT1):c.751+2T>C	ACCGCGGGYAGGGACCTGTGGGG	Diarrhea 7
111033830	NM_000155.3(GALT):c.574A>G (p.Ser192Gly)	TGCYGGGCCATACCTGTCAAGGG, CTGCYGGCCCATACTGTCAAGG	Deficiency of UDPglucose-hexose-1-phosphate uridyltransferase
28933679	NM_000132.3(F8):c.5600A>G (p.His1867Arg)	GAGYGCACATCTTTCTCTAGGG, TGAGYGCACATCTTTCTCTAGG	Hereditary factor VIII deficiency disease
137852251	NM_000133.3(F9):c.917A>G (p.Asn306Ser)	GCTGCAYTGTAGTTGTGGTGAGG	Hereditary factor IX deficiency disease
141686175	NM_001287223.1(SCN11A):c.3473T>C (p.Leu1158Pro)	CGTGCCTCYGTCCCAGTTGAAGG	Episodic pain syndrome, familial, 3
137852331	NM_000402.4(G6PD):c.583A>G (p.Asn195Asp)	ATGCGGTYCCAGCCTCTGCTGGGG	Favism, susceptibility to, Anemia, nonspherocytic hemolytic, due to G6PD deficiency
137852369	NM_000132.3(F8):c.5821A>G (p.Asn1941Asp)	TAGCCATYGATTGCTGGAGAAGG	Hereditary factor VIII deficiency disease
137852389	NM_000132.3(F8):c.398A>G (p.Tyr133Cys)	TCAYATTCAGCTCCTATAGCAGG	Hereditary factor VIII deficiency disease
137852406	NM_000132.3(F8):c.940A>G (p.Thr314Ala)	TGAGCAGYAAGGAAAGTTATTGG	Hereditary factor VIII deficiency disease
28931576	NM_000041.3(APOE):c.178A>G (p.Thr60Ala)	ACAGTGYCTGCACCCAGCGCAGG	
74315301	NM_000396.3(CTSK):c.990A>G (p.Ter330Trp)	GAGYCACATCTGGGGAGCTGG	Pyknodysostosis
137852540	NM_002764.3(PRPS1):c.341A>G (p.Asn114Ser)	TAGCATAYTTGCAACAAGCTTGG	Phosphoribosylpyrophosphate synthetase superactivity
137852624	NM_000215.3(JAK3):c.299A>G (p.Tyr100Cys)	AATCCTGYACAGCAGGACTTGGG	Severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-positive, NK cell-negative
137852640	NM_001166107.1(HMGCS2):c.500A>G (p.Tyr167Cys)	ACCACCGYAGCAGGCATTGGTGG	mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency
137852814	NM_005633.3(SOS1):c.1654A>G (p.Arg552Gly)	GCATCCYTTCCAGTGTACTCCGG	Noonan syndrome, Noonan syndrome 4, Rasopathy
137852865	NM_001171993.1(HPD):c.362A>G (p.Tyr121Cys)	CCTCAYATCCAGGCAAGAATTGG	4-Hydroxyphenylpyruvate dioxygenase deficiency
370898981	NM_138691.2(TMC1):c.1763+3A>G	TGGCCYACCAGATCATGCCTTGG	Deafness, autosomal recessive 7
118192167	NM_000540.2(RYR1):c.14387A>G (p.Tyr4796Cys)	CCATAYACCAGCCCAGGTACAGG	Malignant hyperthermia susceptibility type 1, Central core disease
137852972	NM_032667.6(BSCL2):c.263A>G (p.Asn88Ser)	CGAGACAYTGGCACACAGGGAGG	Distal hereditary motor neuropathy type 5, Silver spastic paraplegia syndrome, Charcot-Marie-Tooth disease, type 2
118192193	NM_172107.2(KCNQ2):c.356A>G (p.Glu119Gly)	CTTCYCATACTCCTTGATGGTGG, GCTCTTCYCATACTCCTTGATGG	Benign familial neonatal seizures 1
118192201	NM_172107.2(KCNQ2):c.622A>G (p.Met208Val)	GGATCAYCCGCAGAACATGCGAGG	Benign familial neonatal seizures 1
137853027	NM_001080463.1(DYNC2H1):c.9044A>G (p.Asp3015Gly)	ATAYCTCTAATTACATCAGGTGG, AGAATAYCTCTAATTACATCAGG	Short-rib thoracic dysplasia 3 with or without polydactyly
137853197	NM_144573.3(NEXN):c.1955A>G (p.Tyr652Cys)	ATAYACTCTCCTCCATCTTCTGG	Dilated cardiomyopathy 1CC, Cardiomyopathy, not specified
137853203	NM_000476.2(AK1):c.491A>G (p.Tyr164Cys)	TTCTCAYAGAAGGGCGATGACGGG, TTTCTCAYAGAAGGGCGATGACGG	Adenylate kinase deficiency, hemolytic anemia due to
786200859	NM_000308.2(CTSA):c.746+3A>G	TCCCAYACCTGTTCCCCAGAAGG	Galactosialidosis, adult

786200897	NM_003494.3(DYSF):c.1285-2A>G	CAGCYAGAAGACACAGGGAGGG, ACAGCYAGAAGACACAGGGAGGG, CACAGCYAGAAGACACAGGGAGG	Limb-girdle muscular dystrophy, type 2B
786200928	NM_206933.2(USH2A):c.7595-2144A>G	CTCTTAYCTTGGAAAGGAGAGG	Usher syndrome, type 2A
137853322	NM_003639.4(IKBKG):c.1219A>G (p.Met407Val)	CCAYATCAGGGGCCTGATACTGG	Incontinentia pigmenti syndrome
387906267	NM_000022.2(ADA):c.219-2A>G	CCCCYGGGAAGGGAAAGAAAGGG, GCCCCYGGGAAGGGAAAGAAAGGG, AGCCCCYGGGAAGGGAAAGAAAGG	Severe combined immunodeficiency due to ADA deficiency
387906362	NM_000492.3(CFTR):c.3717+4A>G	TCAAATCYCACCCCTCTGGCCAGG	Cystic fibrosis
397507442	NM_002769.4(PRSS1):c.65A>G (p.Asp22Gly)	CTTGYCATCATCATCAAAGGGGG, TCTTGYCATTTCATCAAAGGGGG, ATCTTGYCATCATCATCAAAGGG, GATCTTGYCATCATCATCAAAGG	Hereditary pancreatitis
137853971	NM_024598.3(USB1):c.502A>G (p.Arg168Gly)	CCACCYGGTTTCTTGTGATTGG	Poikiloderma with neutropenia
2228063	NM_000067.2(CA2):c.754A>G (p.Asn252Asp)	TGTYCTTCAGTGGCTGAGCTGGG, CTGTYCTTCAGTGGCTGAGCTGG	
387906743	NM_001376.4(DYNC1H1):c.2909A>G (p.Tyr970Cys)	ATTCAAGYAGATTACCTGATTGG	Spinal muscular atrophy, lower extremity predominant 1, autosomal dominant
387906772	NM_002052.4(GATA4):c.928A>G (p.Met310Val)	TCCGCAYTGCAAGAGGCCCTGGG, TTCCGCAYTGCAAGAGGCCCTGGG	Atrial septal defect 2
387906825	NM_000414.3(HSD17B4):c.650A>G (p.Tyr217Cys)	TGCCACAYACTCTGGCTTCAGGG	Gonadal dysgenesis with auditory dysfunction, autosomal recessive inheritance
387906895	NM_006587.3(CORIN):c.1414A>G (p.Ser472Gly)	GGATAACYTGTACTGTTGTAGGG	Preeclampsia/eclampsia 5
387906957	NM_016013.3(NDUFAF1):c.758A>G (p.Lys253Arg)	ACCYTGACCTCCTGCCAGTAGGG, TACCYTGACCTCCTGCCAGTAGG	Mitochondrial complex I deficiency
28933682	NM_000132.3(F8):c.5822A>G (p.Asn1941Ser)	TAGCCAYTGATTGCTGGAGAAGG	Hereditary factor VIII deficiency disease
387907135	NM_016464.4(TMEM138):c.389A>G (p.Tyr130Cys)	CAGYACAACACTGCTGCTGTGGG, GCAGYACAACACTGCTGCTGTGG	Joubert syndrome 16
137854530	NM_001077488.3(GNAS):c.1A>G (p.Met1Val)	GCCCAYGGCGGGCGGGCGGGCGG	Pseudohypoparathyroidism type 1A
387907176	NM_018105.2(THAP1):c.70A>G (p.Lys24Glu)	CCTCACTYGTGAAAGAACAGGG	Dystonia 6, torsion
137854593	NM_000397.3(CYBB):c.1499A>G (p.Asp500Gly)	TCACAYCTTCTCCTCATCATGG	Chronic granulomatous disease, X-linked
387907226	NM_000076.2(CDKN1C):c.832A>G (p.Lys278Glu)	CGCTYGGCGAAGAAATCTGCGGG, GCGCTYGGCGAAGAAATCTGCGG	Intrauterine growth retardation, metaphyseal dysplasia, adrenal hypoplasia congenita, and genital anomalies
387907242	NM_022912.2(REEP1):c.304-2A>G	TCCYGTCAAAGGAAAAACAGAGG	Distal hereditary motor neuropathy type 5B
387907291	NM_022787.3(NMNAT1):c.817A>G (p.Asn273Asp)	TGTYTCTCTGCAAAGGGGCCAGG	Leber congenital amaurosis 9
387907576	NM_001287.5(CLCN7):c.296A>G (p.Tyr99Cys)	TGTCAYAGTCCAAGCTCTGCAGG	Osteopetrosis autosomal dominant type 2, Osteopetrosis autosomal recessive 4

SUPPLEMENTARY REFERENCES

- 35 Maruyama, T. *et al.* Increasing the efficiency of precise genome editing with CRISPR-Cas9 by inhibition of nonhomologous end joining. *Nature biotechnology* **33**, 538-542, doi:10.1038/nbt.3190 (2015).
- 36 Chu, V. T. *et al.* Increasing the efficiency of homology-directed repair for CRISPR-Cas9-induced precise gene editing in mammalian cells. *Nature biotechnology* **33**, 543-548, doi:10.1038/nbt.3198 (2015).
- 37 Lin, S., Staahl, B. T., Alla, R. K. & Doudna, J. A. Enhanced homology-directed human genome engineering by controlled timing of CRISPR/Cas9 delivery. *eLife* **3**, e04766, doi:10.7554/eLife.04766 (2014).
- 38 Ran, F. A. *et al.* Genome engineering using the CRISPR-Cas9 system. *Nat. Protocols* **8**, 2281-2308, doi:10.1038/nprot.2013.143 (2013).
- 39 Richardson, C. D., Ray, G. J., DeWitt, M. A., Curie, G. L. & Corn, J. E. Enhancing homology-directed genome editing by catalytically active and inactive CRISPR-Cas9 using asymmetric donor DNA. *Nat Biotech* **34**, 339-344, doi:10.1038/nbt.3481 (2016).
- 40 Harris, R. S., Petersen-Mahrt, S. K. & Neuberger, M. S. RNA Editing Enzyme APOBEC1 and Some of Its Homologs Can Act as DNA Mutators. *Molecular Cell* **10**, 1247-1253 (2002).
- 41 Cong, L. *et al.* Multiplex genome engineering using CRISPR/Cas systems. *Science* **339**, 819-823 (2013).
- 42 Simonelli, V., Narciso, L., Dogliotti, E. & Fortini, P. Base excision repair intermediates are mutagenic in mammalian cells. *Nucleic acids research* **33**, 4404-4411, doi:10.1093/nar/gki749 (2005).
- 43 Barnes, D. E. & Lindahl, T. Repair and Genetic Consequences of Endogenous DNA Base Damage in Mammalian Cells. *Annual Review of Genetics* **38**, 445-476, doi:doi:10.1146/annurev.genet.38.072902.092448 (2004).
- 44 Tsai, S. Q. *et al.* GUIDE-seq enables genome-wide profiling of off-target cleavage by CRISPR-Cas nucleases. *Nature biotechnology* **33**, 187-197, doi:10.1038/nbt.3117 (2015).
- 45 Ran, F. A. *et al.* In vivo genome editing using *Staphylococcus aureus* Cas9. *Nature* **520**, 186-191, doi:10.1038/nature14299 (2015).
- 46 Kuscu, C., Arslan, S., Singh, R., Thorpe, J. & Adli, M. Genome-wide analysis reveals characteristics of off-target sites bound by the Cas9 endonuclease. *Nature biotechnology* **32**, 677-683, doi:10.1038/nbt.2916 (2014).
- 47 Wu, X. *et al.* Genome-wide binding of the CRISPR endonuclease Cas9 in mammalian cells. *Nature biotechnology* **32**, 670-676, doi:10.1038/nbt.2889 (2014).
- 48 Landrum, M. J. *et al.* ClinVar: public archive of interpretations of clinically relevant variants. *Nucleic acids research*, doi:10.1093/nar/gkv1222 (2015).